THE OPEN UNIVERSITY
SCIENCE: A FOUNDATION COURSE

UNIT 20 INHERITANCE AND CELL DIVISION

EGG

POLLEN

CROSS-FERTILIZATION

STUDY GUIDE

This Unit has several components: the text, a TV programme, two experiments and two AV sequences. The TV programme relates to all the main themes of the Unit, so you are advised to read to the end of the Unit before viewing the programme. The experiments and AV sequences, however, are each closely linked to a particular Section and should be studied at the points in the Unit noted below.

Experiments 1 and 2 each take about 30 minutes and relate to Sections 2.1 and 5.1, respectively. For these you will need the maize cobs, A and B, from your Experiment Kit. The two AV sequences, one in Section 3.2 and the other in Section 6.1, should take 30 minutes and 20 minutes (respectively) to work through.

I INTRODUCTION: DNA AND INHERITANCE

This Unit pursues two key topics that emerge from Unit 19. The first is inheritance, the second is DNA. As you discovered in Unit 19, natural selection can have evolutionary consequences only on characters that are inherited. But how does inheritance work? In what ways do offspring resemble their parents, and what causes such resemblances? As you also discovered in Unit 19, DNA plays a fundamental role in the lives of animals and plants. It can replicate, it is responsible for the synthesis of other chemicals and it is involved in some way in reproduction. But how is DNA involved in reproduction? What part does DNA play in the passing on of characters from one generation to the next?

In this Unit, you will follow two topics side by side. First, in Section 2, you will look at characters of animals and plants, such as their colour or height, and discover the rules that govern their inheritance. Next, in Section 3, you will look at what is going on in the cells of these animals and plants when they reproduce—paying particular attention to what is happening to their DNA. In Section 4, these two approaches are combined when we see how far events within cells can *explain* the patterns of inheritance that you have seen. Sections 5 and 6 then develop the story further.

To understand inheritance you have to operate at two levels: at the level of whole animals or plants, looking at their phenotypes in successive generations; and at the level of the cell, looking at what happens to the DNA during reproduction. In this Unit we shall, to some extent, be jumping backwards and forwards between these two levels, and in so doing we shall also be jumping from the fundamental work of 19th century biologists to present-day molecular biology. In the next Section, we begin at the first of these levels.

2 THE INHERITANCE OF ONE PAIR OF CONTRASTING CHARACTERS

This Section describes one of the simplest known examples of inheritance—the inheritance of a single pair of contrasting phenotypic characters.

2.1 GRAIN COLOUR IN MAIZE

Maize, or corn, occurs throughout the world as an extremely important commercial plant, and it is partly through the interest of plant breeders that maize has been used so extensively in genetic research. Take from the Experiment Kit (Tray C) the maize cob labelled A. Notice that it consists of hundreds of grains (seeds) arranged in columns. Each of the grains on the cob is the result of a separate fertilization of an **egg** (female gamete). Before fertilization, each maize plant carries hundreds of flowers, but the male flowers are separate from the female ones (Figure 1). The male flowers produce **pollen**, containing the male gametes. Normally in maize, the pollen from one plant does not pollinate eggs from the same plant. Instead, it pollinates the eggs of another maize plant. This leads to the male gametes from one plant fertilizing the female gametes of the other plant, a process called **cross-fertilization**.

Thus the maize cob is the result of several hundred cross-fertilizations, and each grain has the potential to develop into a new, individual plant. Each single cob therefore provides a wealth of information about inheritance in the maize plant.

MALE
FLOWER HEAD
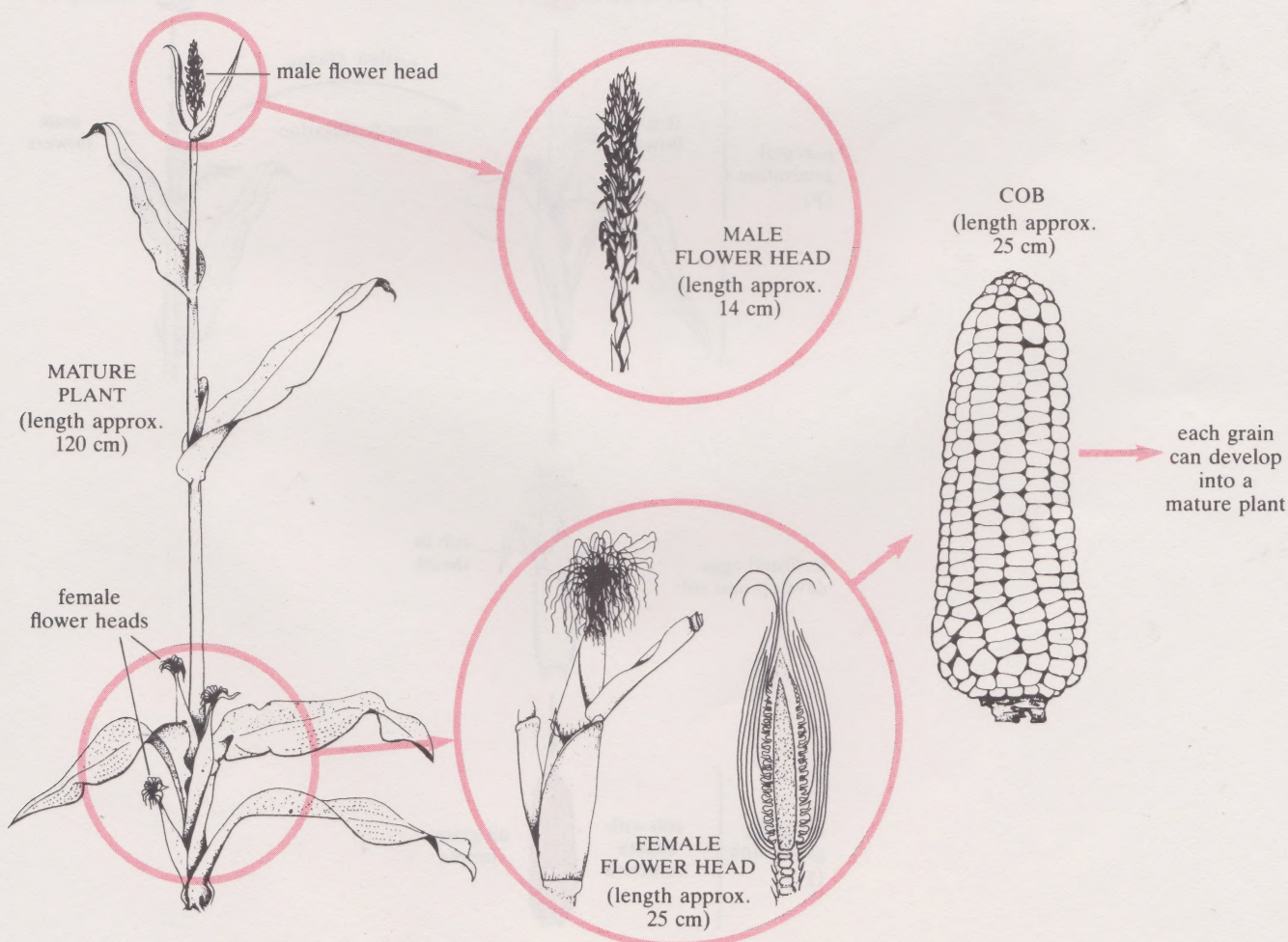(length approx.
14 cm)

COB
(length approx.
25 cm)

MATURE
PLANT
(length approx.
120 cm)

each grain
can develop
into a
mature plant

female
flower heads

FEMALE
FLOWER HEAD
(length approx.
25 cm)

male flower head

FIGURE 1   The life cycle of maize.

The most obvious feature of maize cob A is that the grains are of two colours: some are dark (they are purple in fresh cobs) and some are cream-coloured (nearly white in fresh cobs). Notice that there are no grains of intermediate colour. The two phenotypic characters of the grain are quite distinct: purple and white. Characters that vary in this way are said to show **discontinuous variation**. It turns out that the inheritance of discontinuously varying characters is easier to understand than that of characters that vary continuously (the height of human adults is an example), and for this reason, most of the examples discussed in this Unit are of discontinuous variation.

Each grain is one of two colours; but where is that colour inherited from? The grains on the maize cob in your Kit are the results of a breeding experiment that began two generations earlier, using **pure-breeding** plants. A strain of plants or animals is said to be pure-breeding for a character if all members of that strain have the same character, such as grain colour, and if all breeding within the strain leads to offspring that also have the same character. This particular breeding experiment started with two pure-breeding strains of maize, one strain with purple grains and the other with white grains, and was done in two stages.

In the first stage, shown in Figure 2, plants from the pure-breeding purple-grained strain were cross-fertilized (or crossed, for short) with plants from the pure-breeding white-grained strain. These plants are the **parental generation** (abbreviated to **P**), and the cobs resulting from the cross are the first stage of the offspring generation—even though they are still attached to the parent plant. Another name for this first offspring generation is the **first filial generation** (or **F₁**, for short). Strikingly, the fertilizations resulted in cobs bearing only purple grains. This was true whichever way round the
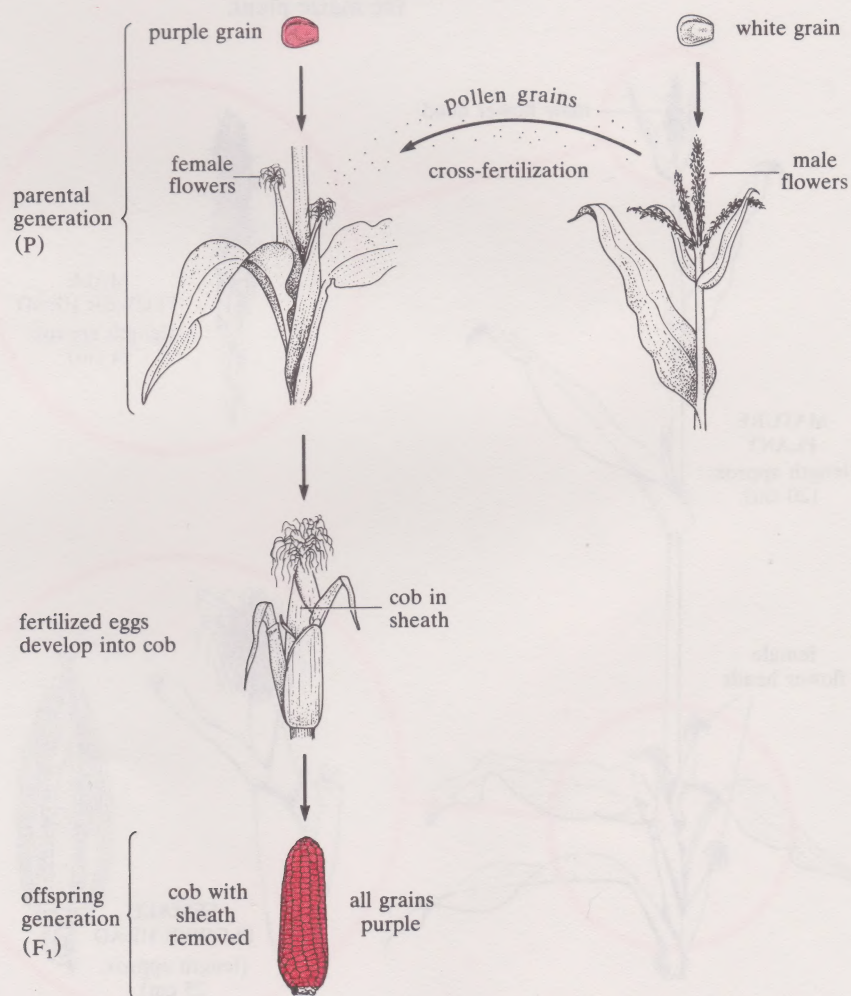

FIGURE 2   The result of crossing plants from a pure-breeding purple-grained strain with plants from a pure-breeding white-grained strain of maize.

purple grain   purple grain

pollen

F₁ adult plants

Leaf-wrapped cob.
The plant bearing the
cob is F₁; the grains on
the cob are the first
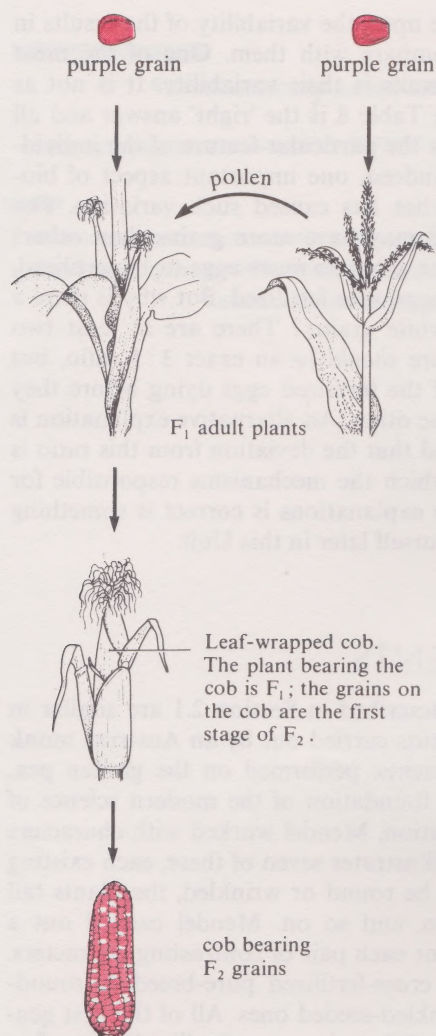stage of F₂.

cob bearing
F₂ grains

FIGURE 3 $F_1 \times F_1$ gives F_2 cobs
(cobs bearing F_2 grains). Cob A in the
Experiment Kit is an F_2 cob.

cross was carried out—that is, regardless of whether the pollen came from the white-grained strain and the eggs from the purple-grained strain, or vice versa.

Two important features of the experiment so far are worthy of emphasis.

1 None of the fertilizations produced white grains. Hence one of the two characters present in the parental generations has vanished in the offspring generation: the offspring of the white-grained plants are not white-grained. Any successful theory of inheritance has to explain this vanishing effect—to explain those cases where parents and offspring differ from rather than resemble one another.

2 The outcome of the breeding experiment did not depend upon which strain donated the eggs and which the pollen. Eggs and pollen must, therefore, make equal contributions to the inheritance of grain colour. Any successful theory of inheritance must account for this equal contribution.

The second stage of the breeding experiment (shown in Figure 3) continued by planting purple grains taken from the cob produced as described above—that is, by planting grains of the F_1 generation. When these had developed into mature plants, they produced male and female flowers which were then cross-fertilized with each other. The fertilized eggs developed into grains borne on cobs. These cobs are the first stage of the second offspring generation—also known as the second filial generation (F_2).

Cob A, which you will use in Experiment 1 below, is an example of an F_2 cob. *You should do this experiment now.*

ITQ 1 How do the colours of the grains in cob A (F_2) compare with those in the previous generation (F_1)? What does this suggest about the factors responsible for their inheritance?

ITQ 2 Use your results to calculate the ratio of purple grains to white grains in cob A. What is the approximate value of this ratio, expressed to the nearest whole numbers?

As you see from the answer to ITQ 2, we looked at a number of different cobs of this type. Table 8 (in the ITQ answer) summarizes our results. Two important points emerge.

1 The total number of grains on a cob can vary greatly—from 348 to 702 in the cobs we looked at.

2 The ratio of purple to white grains can vary a lot too—from 2.52 : 1 to 3.39 : 1 in Table 8. However, all of these ratios are in the region of 3 : 1.

You should remember this ratio because it is of fundamental importance in genetics.

EXPERIMENT 1

THE INHERITANCE OF GRAIN COLOUR IN MAIZE

TIME
This experiment takes about 30 minutes.

NON-KIT ITEM
Felt-tip pen or sticky paper

KIT ITEM
Tray C
Cob A

METHOD

The cob is covered in a plastic coating for protection. Do not remove this covering at any time. Examine the cob and note the colour of the grains. At one end of the cob, lightly mark a point on the plastic cover with either a felt-tip pen or a small piece of sticky paper. This indicates the place where you start counting grains on the cob. Count how many grains there are of each colour on the cob and record your results in your Notebook. When you have finished, remove any mark that you made, taking care not to damage the plastic cover.

For the moment, however, concentrate upon the variability of the results in Table 8 and on how your results compare with them. One of the most characteristic features of biological results is their variability. It is not as though one particular set of figures in Table 8 is the 'right' answer and all the rest are 'mistakes'. Each set reflects the particular feature of the individual cob from which they are taken. Indeed, one important aspect of biological investigation is to find out what has caused such variation. For example, some of the cobs in Table 8 may have more grains than others because there were more flowers on the plant so more eggs to be fertilized, or because a larger proportion of the eggs were fertilized. But why is there a variation in the ratio of purple to white grains? There are at least two possible explanations. One is that there *should be* an exact 3 : 1 ratio, but that this ratio is distorted by some of the fertilized eggs dying before they develop into grains of one colour or the other. An alternative explanation is that there *is not* an exact 3 : 1 ratio and that the deviation from this ratio is a direct consequence of the way in which the mechanisms responsible for inheritance work. Which of these two explanations is correct is something that you will be asked to decide for yourself later in this Unit.

2.2 MENDEL'S EXPERIMENTS

The breeding experiments on maize described in Section 2.1 are similar in essence to a famous series of experiments carried out by an Austrian monk called Gregor Mendel. These experiments, performed on the garden pea, were published in 1865, and laid the foundation of the modern science of genetics. Just as in the maize investigation, Mendel worked with characters that varied discontinuously. Plate 11 illustrates seven of these, each existing as a contrasting pair—the seeds may be round or wrinkled, the plants tall or short, the flowers purple or white, and so on. Mendel carried out a separate set of breeding experiments for each pair of contrasting characters. In one experiment, for example, he cross-fertilized pure-breeding round-seeded plants with pure-breeding wrinkled-seeded ones. All of the first generation offspring were round-seeded. When he cross-fertilized these first generation offspring (F_1) and produced the second generation of offspring (F_2), he found that the latter contained nearly three times as many round-seeded plants as wrinkled-seeded. Table 1 gives the exact numbers, together with the results of six more of Mendel's experiments.

TABLE 1 The numbers of F_2 offspring possessing particular phenotypic characters in seven breeding experiments carried out by Mendel.
The character in italics (and depicted in colour in Plate 11) is the one that is found in all of the F_1 generation of offspring.

Experiment no.	Character	Numbers	Ratio
1	seed shape: *round* or wrinkled	5 474 : 1 850	2.96 : 1
2	seed colour: *yellow* or green	6 022 : 2 001	3.01 : 1
3	pod shape: *inflated* or constricted	882 : 299	2.95 : 1
4	pod colour: *green* or yellow	428 : 152	2.82 : 1
5	flower colour: *purple* or white	705 : 224	3.15 : 1
6	flower position: *along stem* or at tip	651 : 207	3.14 : 1
7	stem length: *long* or short	787 : 277	2.84 : 1

The results in Table 1 are a striking confirmation of what you found with maize in Experiment 1. All the ratios that Mendel found in the F_2 generation were close to, but not exactly equal to, $3:1$. Again, as with maize, the character found in approximately three-quarters of the second generation plants was the one that occurred in all of the first generation plants, and the character found in approximately one-quarter of the second generation plants was the one that had vanished in the first generation of offspring. It is clear then that the maize results discussed in Section 2.1 are not an isolated phenomenon.

The question that now arises is: How can we *explain* Mendel's results? What is it inside cells that produces both these impressively consistent ratios and the other features you observed in the breeding experiments? In fact, it was only around the time of Mendel's death in 1884 that the first fruits of cytology—the study of cell structure—began to offer some clues. The following Section looks at events that occur within cells during the formation of gametes and in fertilization. From that, in Section 4, we hope to find our explanations.

SUMMARY OF SECTION 2

The Section describes the results of breeding experiments involving discontinuously varying characters in maize and garden peas. The main results of these experiments are as follows.

1   It is possible to identify strains that are pure-breeding for a given character. These often form the parental generation (P) in breeding experiments.

2   When plants that differ in the particular character for which they are pure-breeding are crossed, only one of these parental characters appears in the first offspring generation (F_1). The other vanishes. Also, there are no offspring intermediate between the two parents for the particular character.

3   When plants that differ in the particular character for which they are pure-breeding are crossed, the outcome is independent of which parent provides the eggs and which the pollen. Thus the genetic contribution of both parents is equal.

4   When the offspring of parents that differ in the particular character for which they are pure-breeding are themselves crossed, the generation of offspring that they in turn produce contains both of the characters that were present in the original (parental) generation. The character that had vanished in the first offspring generation (F_1) is present in about one-quarter of the individuals in the second offspring generation (F_2). The character that was shown in all of the first offspring generation is shown in about three-quarters of the second offspring generation.

5   Although the ratio of the numbers of individuals in the second offspring generation with the respective two characters is about $3:1$, there is variation in this ratio, for it is rarely exactly $3:1$.

SAQ I   Which of the following breeding experiments could have involved pure-breeding parents? Give reasons for your choice, including reasons for rejecting the options you do not choose. (Judge each option separately from the others.)

(Option 1)   300 white-eyed flies are crossed with 300 red-eyed flies. All of the offspring are red-eyed.

(Option 2)   300 white-eyed flies are crossed with 300 red-eyed flies. Half of the offspring are white-eyed and half are red-eyed.

(Option 3)   300 white-eyed flies are crossed with 300 white-eyed flies. Half of the offspring are white-eyed and half are red-eyed.

(Option 4)   300 white-eyed flies are crossed with 300 white-eyed flies. All of the offspring are white-eyed.

7

SPERM

OVUM

OVULE

CHROMOSOMES

HISTONES

SAQ 2 In what respect does each of the following imaginary breeding experiments produce results that differ in an important way from the results obtained by Mendel?

Experiment 1 100 pure-breeding red-flowered plants are crossed with 100 pure-breeding white-flowered plants. All of the offspring have red flowers. These offspring are then crossed with each other. About three-quarters of their offspring have white flowers, and about one-quarter have red flowers.

Experiment 2 100 pure-breeding red-flowered plants are crossed with 100 pure-breeding white-flowered plants. All of the offspring have pink flowers.

Experiment 3 When the pollen from 100 pure-breeding red-flowered plants pollinates the eggs of 100 pure-breeding white-flowered plants all of the offspring have white flowers. When the pollen from 100 pure-breeding white-flowered plants pollinates the eggs of 100 pure-breeding red-flowered plants all of the offspring have red flowers.

3 MEIOSIS

In the previous Section, we explored the pattern of inheritance of particular characters. In this Section, we examine the events that occur within cells, both during the formation of gametes and at fertilization, in order to see how Mendel's results can be explained.

This Section covers about a hundred years of discovery in the fields of cytology and, more recently, molecular biology. The approximate beginning of this period was the first sighting, by Walther Flemming in 1882, of threadlike bodies in the nuclei of dividing salamander cells: the first observation of chromosomes. Further cytological studies advanced knowledge of chromosome behaviour, and finally—with increasing of knowledge of DNA and its role in genetics and chromosome structure—we reach the present time.

3.1 DNA AND CHROMOSOMES

In Unit 19, you read that the bodies of living organisms are made up of cells, and that in eukaryotes (that is, organisms whose cells have nuclei) the DNA is contained within the nucleus. You also learned that organisms reproduce by means of simple, single-celled structures called gametes, and that sexual reproduction involves the fusion of two gametes (that is, fertilization). To understand the inheritance of the characters described in the previous Section it is crucial to know what the DNA is like in the cells of the sexually-reproducing parents, and also what happens to it when they produce gametes and when the gametes fuse at fertilization.

In animals, the male gamete is the **sperm** (or spermatazoon, plural spermatozoa) and the female gamete is the egg or **ovum**. In plants, the male gamete (which develops from the pollen grain) is also called the sperm and the female gamete is the egg or **ovule***. In animals sperms are typically very small (see Figure 4), consisting of a head, containing the nucleus, and a tail, and are produced in vast numbers. A single male human ejaculation releases hundreds of millions of sperms. Females produce far fewer gametes than do males of their species—a woman will produce only about 200–500 eggs during her lifetime, but each egg (Figure 5) is very much bigger than a sperm.
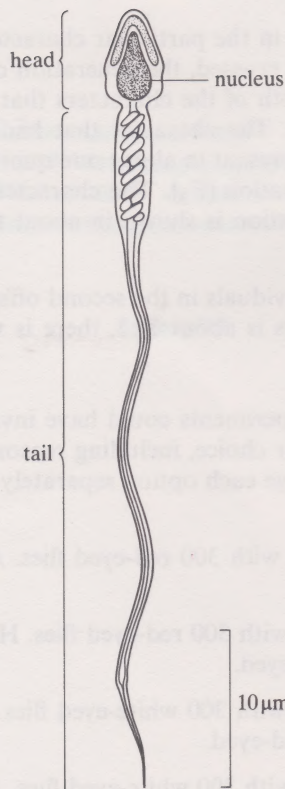

head

nucleus

tail

10 μm

FIGURE 4 A human sperm.

* The situation in plants is rather more complicated than this, but for simplicity we shall use the term 'sperm' for the male gamete and 'ovule' for the female gamete.

typical body cell

nucleus

human egg

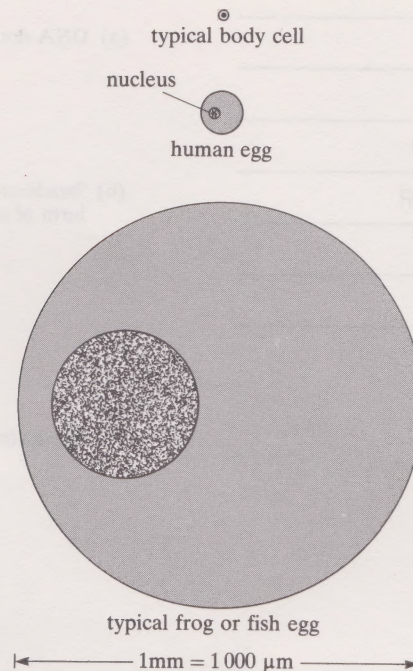typical frog or fish egg

|←——— 1mm = 1 000 µm ———→|

FIGURE 5 The relative sizes of various eggs compared with that of a typical human body cell.

DNA is an important component of structures inside the nucleus called **chromosomes**. These are more easily seen under the microscope at some stages in a cell's life history than at others, and are usually most easily visible when the cell is dividing. Typically, each chromosome looks like a rod or thread under the light microscope, but this simple appearance belies an extraordinarily intricate underlying structure. Human chromosomes differ in size (see Figure 6) but estimates suggest that each chromosome consists of a single DNA molecule, around 5 cm long, containing about 1.3×10^8 nucleotides. The nucleus of a human body cell is usually about 5 µm in diameter, and there are 46 chromosomes in the nucleus of a typical body cell (such as a muscle or nerve cell), so packaging the enormously long DNA molecules is an amazing feat. The chromosome also contains many different proteins, and one group of proteins, the **histones**, play an important role in the packaging.
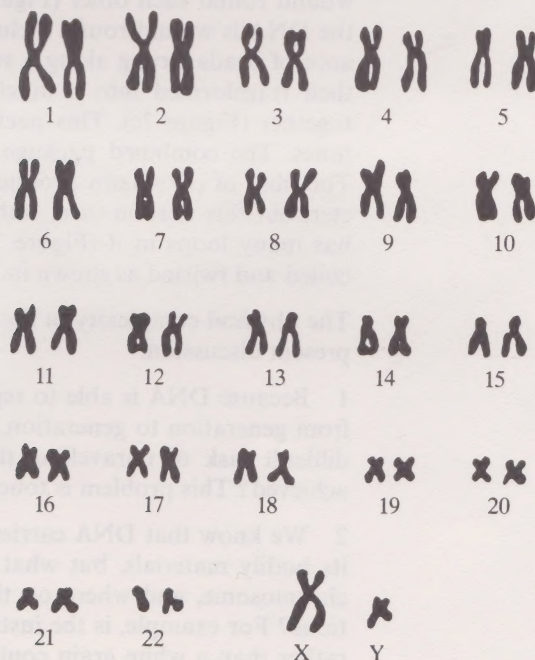
FIGURE 6 Chromosomes from a human male, lined up in pairs. There are 46 chromosomes: 22 matching pairs and the two sex chromosomes (X and Y) that are not identical (the Y is smaller than the X).

CHROMATIN

SOMATIC CELLS

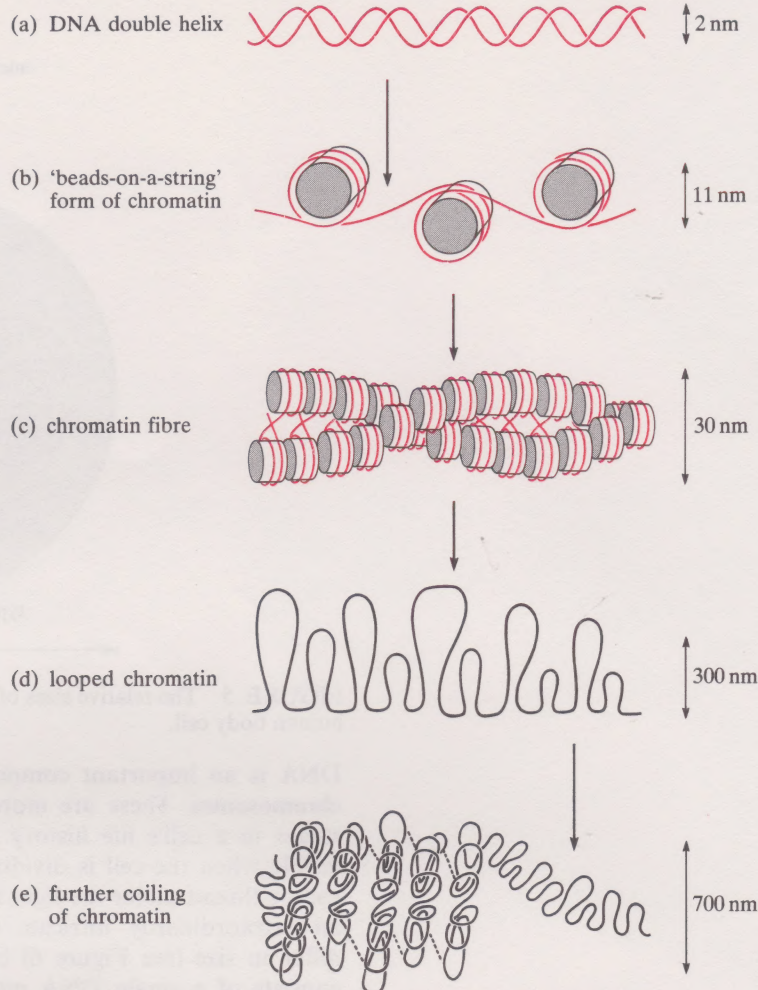DIPLOID NUMBER

HAPLOID NUMBER

ZYGOTE

MEIOSIS

(a) DNA double helix   2 nm

(b) 'beads-on-a-string' form of chromatin   11 nm

(c) chromatin fibre   30 nm

(d) looped chromatin   300 nm

(e) further coiling of chromatin   700 nm

FIGURE 7 Packing arrangements of chromatin within a chromosome ($1\,nm = 10^{-9}\,m$). You need not remember the details of this Figure, but only that a mechanism exists within the cell for packaging the long strands of DNA.

We can imagine the DNA molecule as a piece of string. As you know from Units 17–18, the 'piece of string' consists of two polynucleotide strands wound round each other (Figure 7a). At frequent intervals along its length the DNA is wound round a cluster of histone molecules, giving the appearance of beads strung along a string (Figure 7b). This long, thin structure is then transformed into a much shorter, fatter one by packing the beads together (Figure 7c). This packing is achieved with the help of other histones. The combined package of DNA plus histones is called **chromatin**. The fibre of chromatin produced by this packing is about 30 nm in diameter, but this fibre in turn, rather than being like a straight piece of string, has many loops in it (Figure 7d). These loops may themselves in turn be coiled and twisted as shown in Figure 7e.

The physical complexity of the DNA raises two important problems for the present discussion.

1 Because DNA is able to replicate, with copies of DNA being passed on from generation to generation, this replication must involve the formidably difficult task of unravelling the DNA molecule. How is this unravelling achieved? This problem is touched on again in Unit 24.

2 We know that DNA carries the organism's instructions for synthesizing its bodily materials, but what exactly constitutes 'an instruction' within a chromosome, and where on the chromosome does one find such instructions? For example, is the instruction to a maize plant to produce a purple rather than a white grain contained on one bead of chromatin, or on a bit of DNA between two beads, or on a loop of packed chromatin fibre? Or is there no direct correspondence between 'an instruction' and a physically identifiable part of the chromosome's structure? We return to this problem later in the Unit.

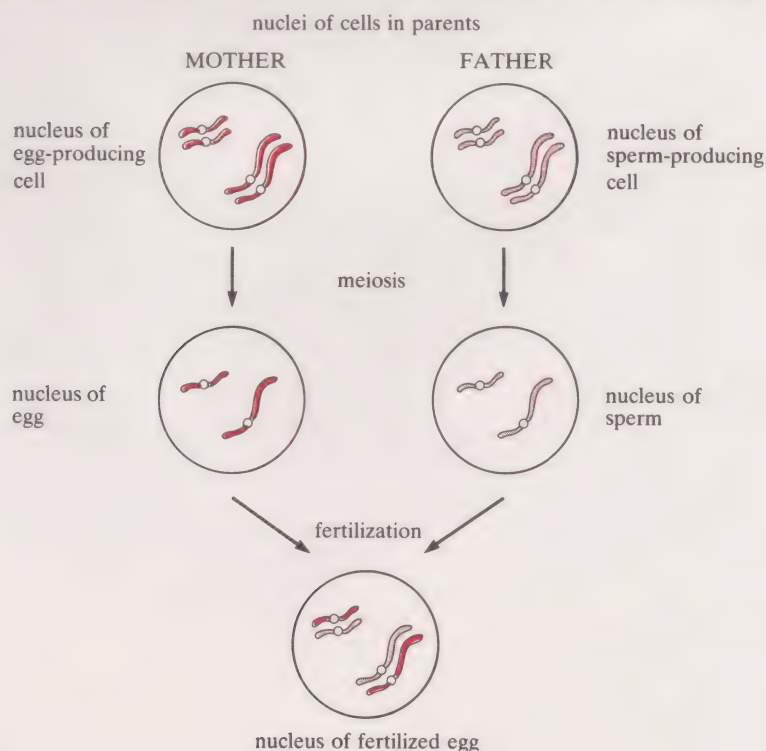## 3.2 CHROMOSOMES AND GAMETE PRODUCTION: AN OUTLINE

Cells that are *not* reproductive cells, i.e. not gametes, are known as **somatic cells**. Your skin cells, muscle cells, nerve cells and so on are all somatic cells: the term is an important one. Significantly, the number of chromosomes in the nucleus of a typical somatic cell of a given animal or plant is always twice the number found in the nucleus of one of its gametes. Thus the nucleus of a human nerve cell or muscle cell, for example, contains 46 chromosomes, whereas the nucleus of a human egg or sperm contains 23. Similarly, each somatic cell of the broad bean plant, *Vicia faba*, contains 12 chromosomes, whereas each egg or sperm produced by it contains but 6.

The number of chromosomes in the non-reproductive cells of an animal or plant is called the **diploid number**. The general term for the number of chromosomes in the gametes of an animal or plant is the **haploid number**. For humans the diploid number of chromosomes is therefore 46 and the haploid number is 23.

In animals and plants, haploid gametes are produced from precursor somatic cells. In men, the sperms (containing 23 chromosomes) are produced from diploid 'sperm mother cells', also called spermatocytes. In women, the ova (also containing 23 chromosomes) are produced from diploid 'egg mother cells', or oocytes. When the two haploid gametes fuse at fertilization, a new diploid cell is produced. This first cell of the new individual is called the **zygote**.

☐ Summarize the changes in chromosome number that occur when a cell gives rise to a gamete, and when gametes produce a fertilized egg, i.e. a zygote.

■ When a cell with the full (diploid) number of chromosomes gives rise to a gamete, the number of chromosomes must somehow be halved. When fertilization happens, the nuclei from the two gametes, each with its half (haploid) set of chromosomes, come together, restoring the number to the diploid value.

Figure 8 shows these changes in chromosome numbers. They occur, not just in humans, but in all animals and plants. The production of a gamete requires a very special process. It involves **meiosis**, a form of cell division in which the number of chromosomes is halved from the diploid to the

FIGURE 8   How the numbers of chromosomes in the nucleus change with gamete production and fertilization. Only two pairs of chromosomes are shown for simplicity.

haploid number. Meiosis, as you will soon discover, holds the key to understanding the pattern of inheritance of different forms of a particular character.

The whole process of meiosis can take days, months or even years to complete, depending on the species of animal or plant involved. Indeed, in women, meiosis can last for up to 50 years—an astonishing state of affairs that we explain in Section 3.3.

Many things happen inside cells undergoing meiosis, but some of the most spectacular and intricate events involve the chromosomes, and it is upon the chromosomes that we are now going to concentrate most of our attention. To help you understand meiosis you should work through the following AV sequence, entitled 'Chromosomes and meiosis', which you will find on Tape 3 (Side 2, Band 3). As well as referring to Frames 1 to 10 during the AV sequence, you will also need to look at Figures 9 and 10. The sequence should take you about 30 minutes to complete.
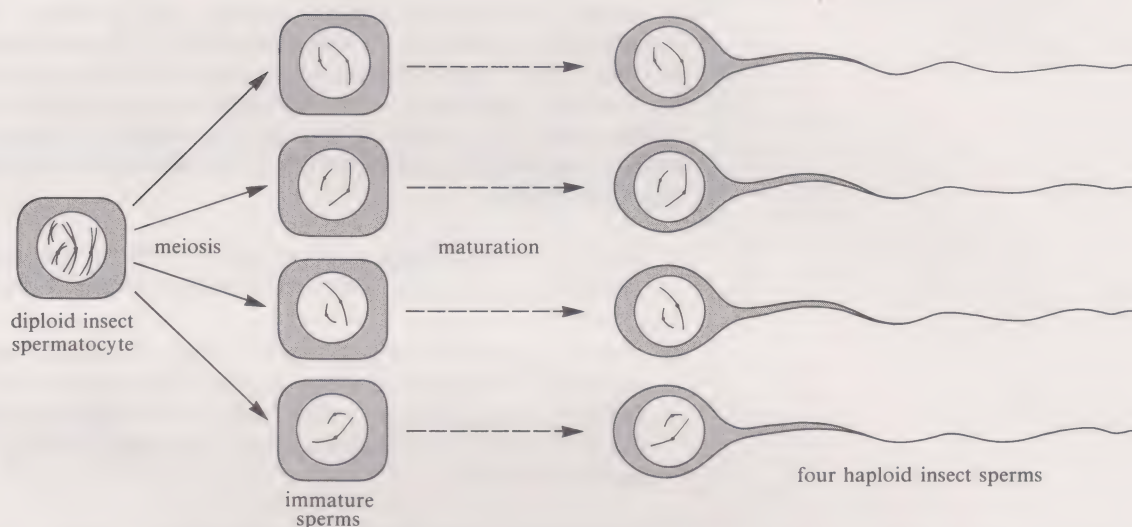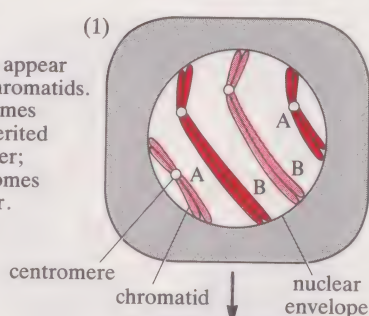

FIGURE 9   Spermatocyte to sperms: the overall accomplishment of meiosis. The four spermatocyte chromosomes are doubled-up structures as shown; you will learn why in the AV sequence 'Chromosomes and meiosis'.
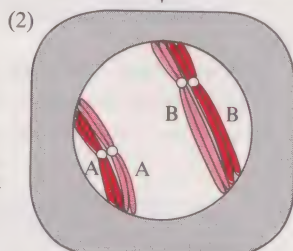
DIVISION I OF MEIOSIS

What happens

(1)

Chromosomes appear divided into chromatids. Red chromosomes have been inherited from the mother; pink chromosomes from the father.
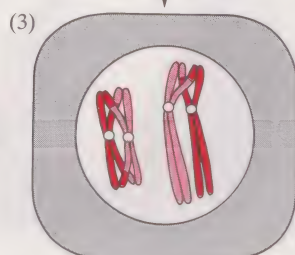
centromere

chromatid

nuclear envelope

Name of phase

prophase I

FIGURE 10 The successive stages of meiosis. The stages have been lined up so that the equivalent phases of division I and division II are side by side. Note how meiosis results in a halving of the number of chromosomes and in the partial exchange of genetic material between homologous chromosomes. The Figure shows meiosis as a process occurring in discrete stages, but it is actually continuous, with one phase leading into the next. Moreover the Figure gives little indication of the relative durations of the phases; in fact prophase I is by far the longest phase (see text later) while anaphase, for example, occupies a very small proportion of the meiotic cycle.

(2)

Chromosomes pair up in homologous pairs. The two chromosomes, A, form one pair, the two chromosomes, B, form another.

prophase I

DIVISION II OF MEIOSIS

What happens

(7)

Beginning of division II. (Only one of the two cells of Figure 10 (6) is shown here— the upper one).

Name of phase

prophase II

(3)

Chromatids cross over and exchange material

prophase I

(8)

Nuclear envelope breaks down and chromosomes align themselves along the middle of the cell.
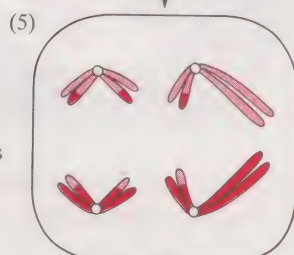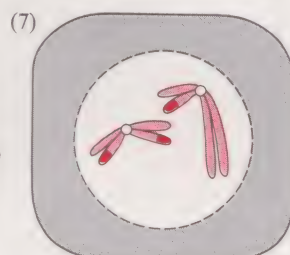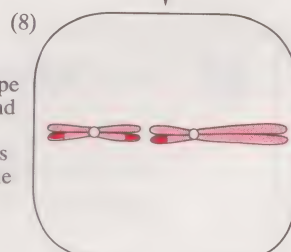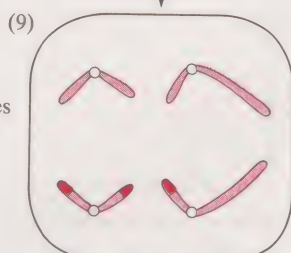
metaphase II

(4)

Nuclear envelope disappears and chromosomes line up around 'equator'.

metaphase I

(5)
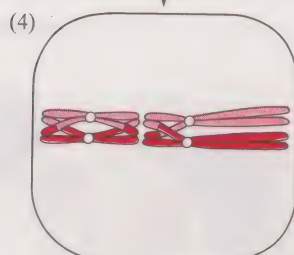
The chromosomes now pull away and move to opposite ends of the cell.

anaphase I

(9)

The centromeres divide and the chromatids move apart.

anaphase II

(6)

Nuclear envelopes develop and cell divides into two.

nuclear envelope

telophase I

(10)

New nuclear envelopes enclose the two collections of chromatids, and the cell divides into two.

nuclear envelope

telophase II

13
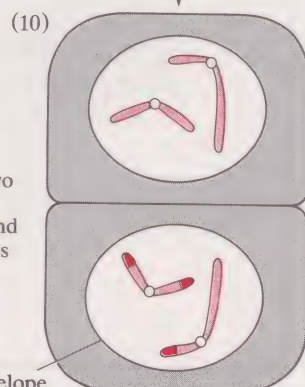
1 Early prophase I
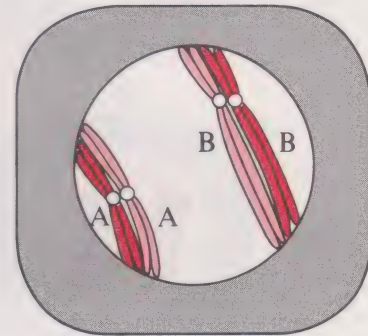
The chromosomes appear
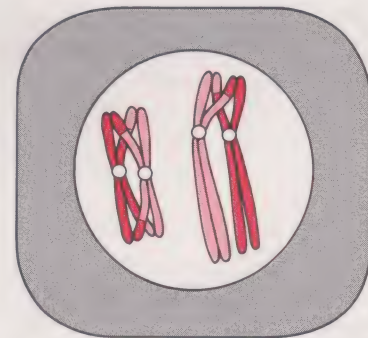
centromere   chromatid   nuclear envelope
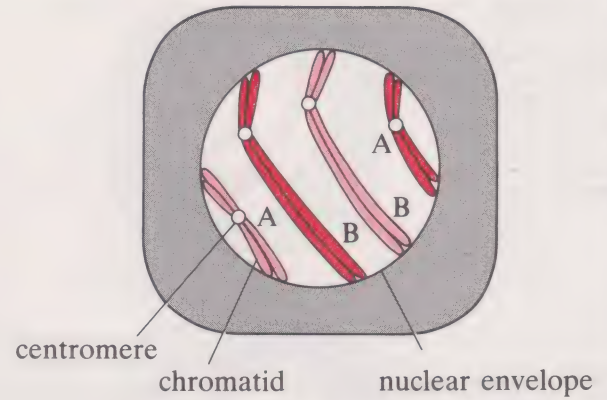
2 Mid-prophase I
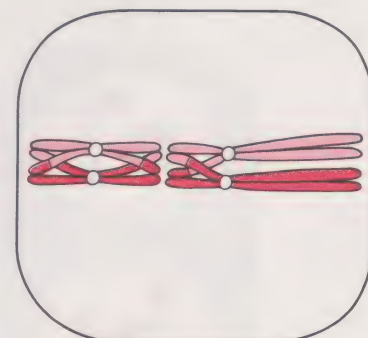
The chromosomes form
pairs

3 Late prophase I
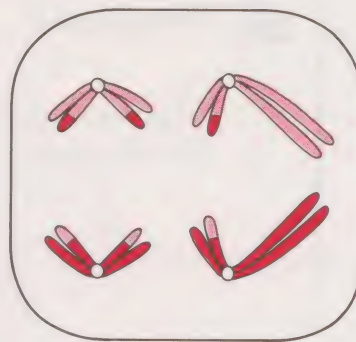
The chromosomes exchange
material

4 Metaphase I

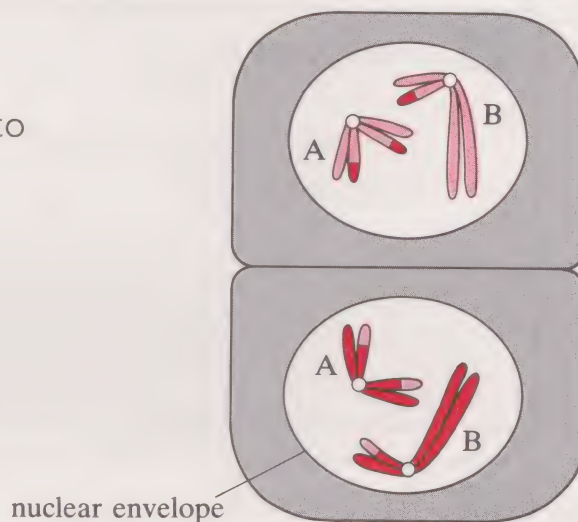The chromosomes line up
along the equator

5 Anaphase I

The chromosomes move apart

6 Telophase I

The cell divides into two

nuclear envelope

Q Some questions on division I of meiosis

The insect spermatocyte contained 4 chromosomes. At the end of meiotic division I, the spermatocyte has given rise to two new cells.

☐ How many pairs of homologous chromosomes are there in the spermatocyte?
....

☐ How many chromatids are there altogether in the spermatocyte?
....

☐ How many pairs of homologous chromosomes are there in each cell produced by division I?
....

☐ How many chromatids are there altogether in each cell produced by division I?
....

☐ Write a sentence summing up how each cell produced by division I compares with the original spermatocyte.

....

....

....

....
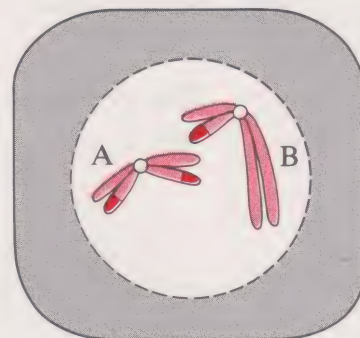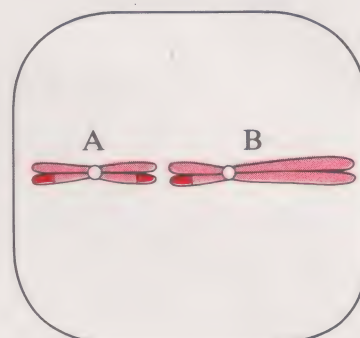
Division II

7 Prophase II
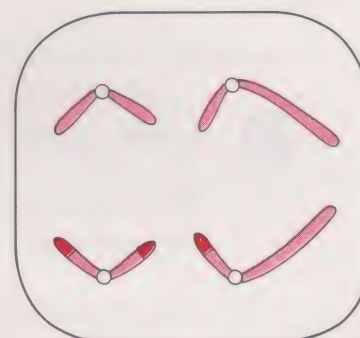
The nuclear envelope
breaks down

8 Metaphase II

The chromosomes line up
along the equator

9 Anaphase II
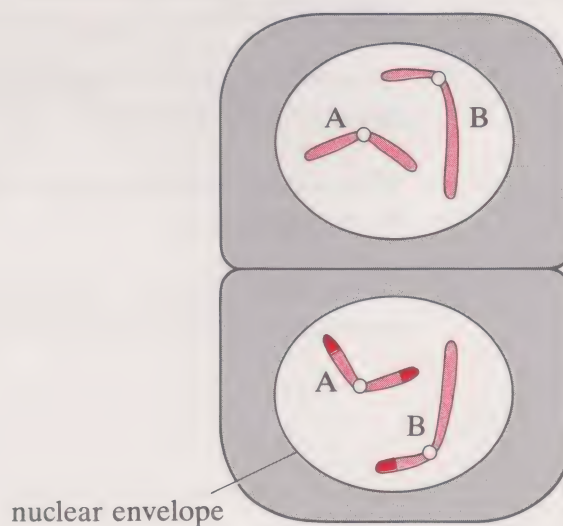
The chromosomes move
apart

10 Telophase II

The cell divides into
two

nuclear envelope

As you learned in the AV sequence, meiosis ends in the production of haploid cells (cells with half the normal ration of chromosomes) and it is these cells, and only these cells, that are suitable to act as gametes. Many of them do so, becoming sperms or eggs with the specialized features such as tails or large size, mentioned earlier. Although there may be thousands of pollen tubes growing down towards the few ovules (eggs) in a single flower, or thousands of sperms clustering round a single animal egg (Figures 11 and 12), only one male gamete will fuse with each female gamete. A sperm

pollen grains
on stigma
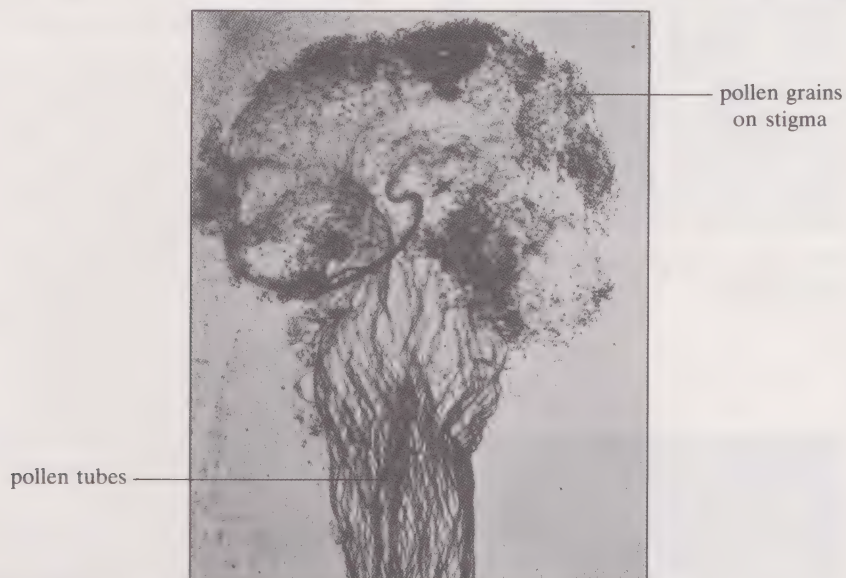
pollen tubes

FIGURE 11   Pollen grains putting out tubes that grow down from the stigma towards the ovules (eggs). The sperm cells in the pollen grains travel down the pollen tubes and fertilize the ovules at the base of the flower.

can bore through the outer layer of an egg, and within a few minutes the nuclei of the two gametes fuse, creating one single diploid nucleus. The zygote then starts to develop. It too divides, first into two, then four, then eight and ultimately into the thousands, millions or billions of somatic cells that make up the mature organism. These cell divisions are *not* meiosis. They are a different form of cell division—called *mitosis*—and this is discussed later in the Unit. The important point for now is that each cell produced by mitosis is diploid, and has a set of chromosomes identical to that of all other somatic cells—including the zygote from which they all come.

50 μm
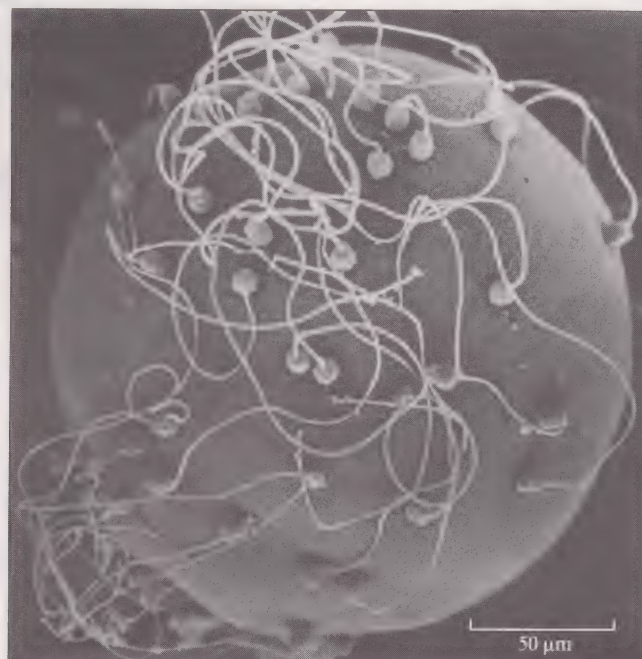
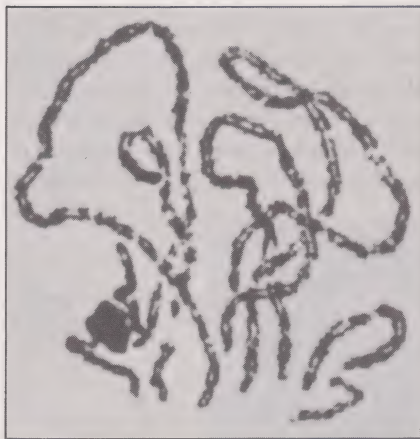FIGURE 12   An egg with many sperms on its surface.
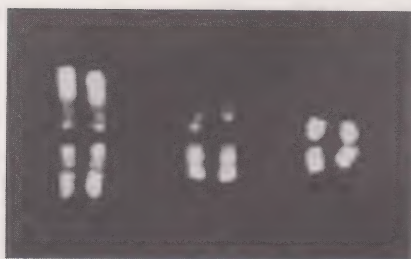
3.3 MEIOSIS: SOME DETAILS

The outline of meiosis presented in Section 3.2 raises some intriguing questions. How do homologous chromosomes recognize each other? What makes them align at the equator and then move apart again? How can we explain the exchange of chromosomal material at crossing over, bearing in mind the complex physical structure of the chromosome described in Section 3.1? It is not possible to answer these questions completely: biological research has not yet progressed that far. However, this Section explains some of the mechanisms that are known to operate during meiosis.

As explained in Section 3.2, a maternally-derived and a paternally-derived chromosome of a homologous pair resemble each other in size, shape and location of the centromere. Figure 13 shows this in the case of a female grasshopper. You may be able to make out eight pairs of chromosomes. Some, like the three pairs in the bottom right-hand corner of Figure 13, are short; others, like the one at the top left, are extremely long. Each member of a homologous pair also shares the same pattern of 'bumps' along its length. The similarity in structure of a homologous pair of chromosomes goes much further than this, however, as becomes clear when one stains the chromosomes with special dyes that fluoresce when they are combined chemically with DNA, and when one examines the chromosomes at a high magnification. Each chromosome then reveals an intricate banding pattern that is shared only by its homologue (that is, the other chromosome of the homologous pair), and not by any of the other chromosomes.

You can also see banding in the three pairs of human chromosomes in Figure 14. It is possible that each band reflects some feature of the physical packing of the loops of chromatin referred to earlier. In one band, the loops might be packed tightly in a particular formation, giving a dense appearance; in a nearby, lighter-coloured band, the packing arrangement may be different, perhaps less dense (Figure 15).

Whatever the explanation, it is clear that the detailed structure of the DNA molecule in each of a pair of homologous chromosomes is strikingly similar. If we think of a chromosome as carrying genetic information, then

FIGURE 13   Eight pairs of grasshopper chromosomes.

FIGURE 14   Three pairs of human chromosomes, stained to show their banding patterns.

loop in chromatin fibre

chromatin fibre

further folding within chromatin loop

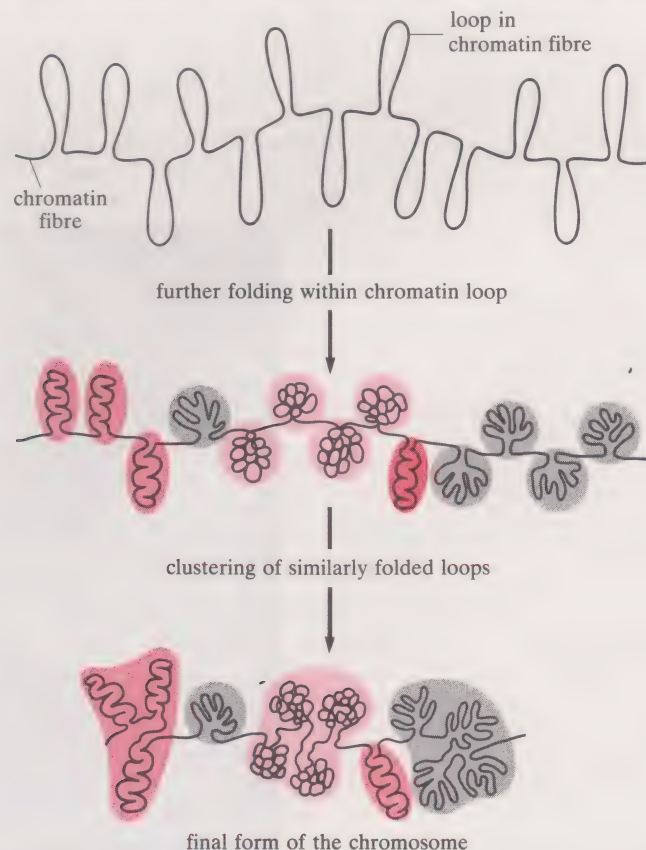clustering of similarly folded loops

final form of the chromosome

FIGURE 15   Showing how the packaging of the chromatin could produce the bands seen on chromosomes.

CHIASMA

the information carried by each member of a homologous pair must have a lot in common.

How homologous chromosomes recognize each other and pair during meiosis is not known. It seems that each chromosome develops a strip of protein along one side, and that the protein strips from a homologous pair, and *only* those from a homologous pair, match perfectly so that they can interlock with one another. At the beginning of meiosis, chromosomes are attached at their ends to the nuclear envelope, and in some cases the pairing process between homologues starts where they join the nuclear envelope, and runs along the chromosomes, like a zip fastener (Figure 16). The result of this process, however it works, is that the two chromosomes of a homologous pair are held together very tightly. They may stay like this for days or even years.
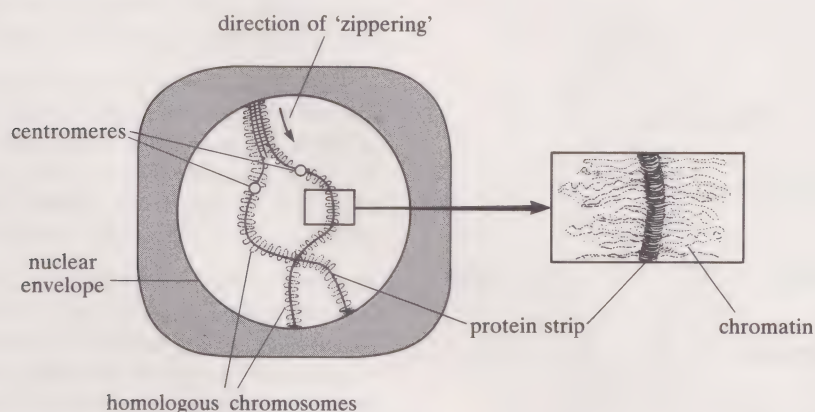
FIGURE 16   The pairing mechanism of homologous chromosomes during meiosis.

FIGURE 17   Crossing over in grasshopper chromosomes during meiosis.

It is interesting that the protein strip comes between the two chromosomes, and so keeps the chromatin of the two chromosomes apart. One might have thought that such an arrangement would have hindered the exchange of chromatin between adjacent chromatids of homologous chromosomes during crossing over, but this does not appear to be so. How exactly the exchange of chromatin between the chromatids is achieved during the time when the homologous chromosomes are tightly bound together is not known. The result, visually, is unmistakable when this period of pairing is over and the homologous chromosomes pull apart. Look at Figure 17, which shows the chromosomes of a grasshopper during late prophase I of meiosis. Examine the pair in the middle at the top (marked A). There are five places at which the chromatids cross over, weaving backwards and forwards from one chromosome to the other. Each point where the chromatids cross over one another is called a **chiasma** (plural **chiasmata**). This pair of chromosomes has five chiasmata, the pair below them (B), has two.

Now look at Figure 18, which is an enlarged tracing of these two sets of chromosomes. It shows diagramatically where the material from one chromosome begins and the other ends. You can see from this diagram that
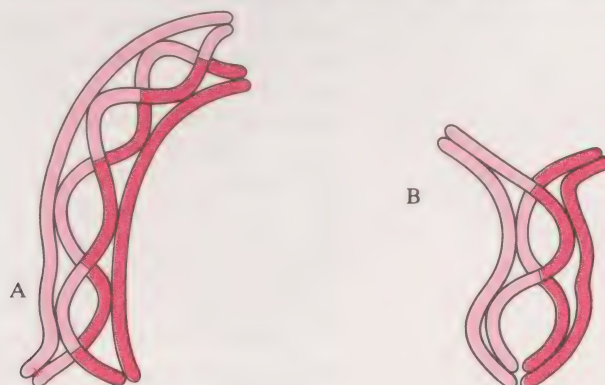
FIGURE 18   Crossing over in the two pairs of chromosomes A and B in Figure 17.
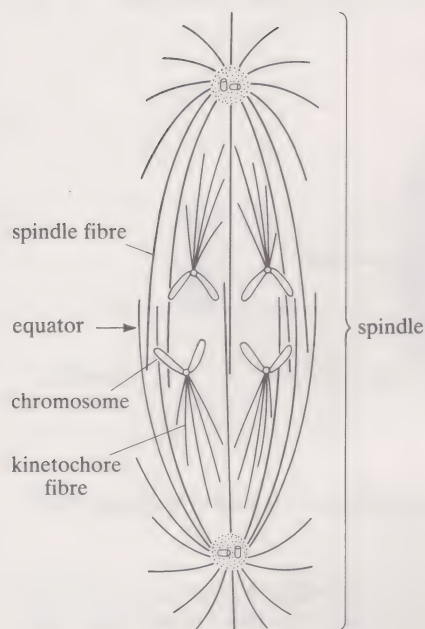
SPINDLE

KARYOTYPE

each crossing over requires a chromatid from each homologous chromosome to break and to join with the broken end of the chromatid from the adjacent chromosome. As Figure 17 shows, crossing over occurs in most pairs of homologous chromosomes during meiosis in the grasshopper. The same is true in humans, where each homologous pair usually has two or three such chiasmata.

The movements of the chromosomes during meiosis are brought about by a system of very fine fibres within the cell, made of a protein called tubulin. There are two sorts of fibres. One sort runs from the 'north' and 'south' poles of the cell towards the equator, like so many lines of longitude on the globe. These are the spindle fibres. The other sort are attached to the chromosomes themselves. They are called kinetochore fibres. Together, these two sorts of fibre make up what is called the **spindle** (Figure 19). The movements of the chromosomes are brought about by these fibres, in a way that will be described in more detail in Section 6.1.

You will recall from the AV sequence that each of the two divisions of meiosis can be subdivided into four named stages. Thus we have, in meiotic division I: prophase I, metaphase I, anaphase I, and telophase I. Similarly, in meiotic division II we have: prophase II, metaphase II, anaphase II, and telophase II. You know already (from the AV sequence) the key features of each stage. The following, however, makes some additional points.

Prophase I lasts from the time when the chromosomes first appear as discrete threads, until after the close contact between the pairs of homologous chromosomes has ended, the chiasmata are clearly visible, and the chromosomes have detached themselves from the nuclear envelope. Prophase I is itself divided into a number of phases, each with its own name, but these are not important here. Prophase I is by far the longest phase of meiosis: it normally occupies about 90% of the whole meiotic cycle. In women it is exceedingly long. Meiosis actually starts (in the oocytes—the precursor cells within the ovaries) while the woman-to-be is still an embryo inside her mother's womb. The whole process comes to a halt before the baby is born at the part of prophase I illustrated in Figure 10, stage 3. Meiosis does not start up again until puberty. Then, during the woman's reproductive life, one cell completes the meiotic division and gives rise to an egg every time she undergoes a menstrual cycle. (Sometimes more than one egg is produced in the same cycle, as evidenced by the occurrence of multiple births.) A cell that completes meiosis shortly after puberty has thus been arrested at prophase I for about 10–14 years. By contrast, a cell that completes meiosis to give rise to an egg late in a woman's reproductive life will have been arrested at prophase I for up to about 50 years.


FIGURE 19   The spindle (at early anaphase).

Such very long durations of prophase I are accompanied by risks of genetic abnormalities. For example, women who give birth after the age of 40 are increasingly likely to have children with Down's syndrome. (Figure 20a shows Cathy, who has Down's syndrome, with her mother Chris.) There are different types of Down's syndrome, but the commonest arises when the child has three copies of chromosome 21 in each somatic cell instead of the normal two. The reason for this genetic abnormality is that, during meiosis leading to the production of the egg, the copies of chromosome 21 fail to separate at anaphase I. Instead, both pairs of chromatids move to the same end of the cell, resulting in an egg containing *two* copies of chromosome 21 instead of one. (An egg produced with *no* chromosome 21 fails entirely.) The sperm contributes an additional chromosome 21, so that the zygote has three copies of the chromosome—with all its consequent effects on the developing child. The reasons why the presence of an extra chromosome has such a profound effect on phenotype are still largely unknown. Figure 20b shows the chromosomes present in each somatic cell of a Down's syndrome girl. (This kind of reconstructed photograph, where the homologous chromosomes are artificially laid out in pairs, shows what is called a **karyotype** of the organism.)

The longer a woman's oocyte is immobilized at prophase I, the greater the chance that chromosomal abnormalities (such as the failure of homologous pairs to separate) will occur. Thus the older a woman is, the greater the
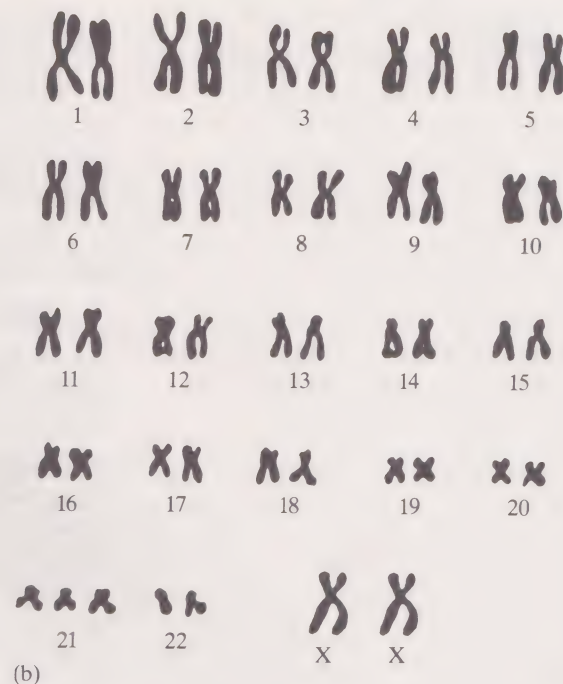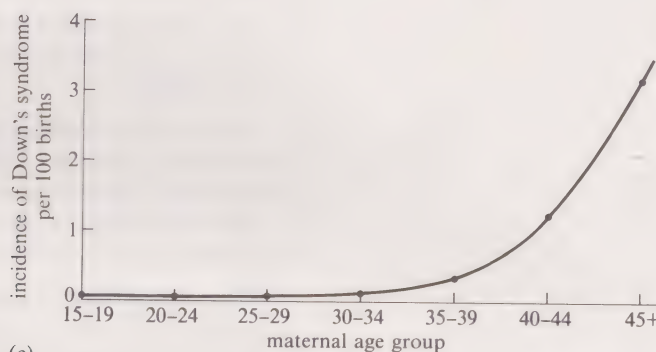
FIGURE 20 Down's syndrome: (a) a mother with her Down's syndrome daughter; (b) the karyotype of a Down's syndrome girl; (c) a graph showing the increase in incidence of Down's syndrome with maternal age.

likelihood that she will produce a defective ovum— having two instead of one copy of chromosome 21. Figure 20c shows how the incidence of Down's syndrome increases with the mother's age. The incidence is very low in younger women but rises steeply to more than 3 per 100 births in mothers aged 45 years and over.

The next stage, after prophase I, where the chromosomes arrange themselves around the equator of the cell, is called metaphase I. The brief stage where the chromosomes pull away to opposite ends of the cell is called anaphase I, and the stage where the chromosomes gather at each end of the cell is called telophase I. The first cell division is then completed. Each cell so produced then enters prophase II. The pairs of chromatids arrange themselves around the equator at metaphase II, pull apart at anaphase II, cluster at opposite ends of the cell at telophase II, and the second cell division is then completed.

SUMMARY OF SECTION 3

There is a lot of detailed factual information in this Section. You should have taken it in and understood it as you read the Section, but we do not expect you to remember it all. However, we do expect you to remember the most important points. These are listed below.

1 Chromosomes contain DNA and histone proteins which are packaged together in a highly convoluted fashion. Replication of DNA therefore involves a complicated unravelling of the coiled chromosomal material.

21

GENE

2   The number of chromosomes in non-reproductive body cells (somatic cells) is called the diploid number. The number of chromosomes in gametes is half of the diploid number, and is called the haploid number.

3   Certain somatic cells give rise to gametes by a process of cell division called meiosis. At the start of meiosis, a cell is diploid. By means of meiosis it gives rise to four haploid cells.

4   When two haploid gametes fuse at fertilization, they give rise to the first cell of the new organism. This is called a zygote.

5   Meiosis consists of two successive cell divisions: division I and division II.

6   By the time that the chromosomes become visible at the beginning of meiosis, each has divided along its length into two identical chromatids. These are held tightly together, and especially securely at the centromere.

7   During division I of meiosis chromosomes come together in homologous pairs, one chromosome from each pair having been inherited from the mother and the other from the father. The two chromosomes belonging to a homologous pair resemble each other closely in physical appearance. This similarity extends to details of the packing arrangement of their DNA.

8   The chromatids from the chromosomes of a homologous pair exchange material in the process called crossing over. Each point along the length of the chromatids where such an exchange of material occurs is called a chiasma (plural chiasmata).

9   The chromosomes of each homologous pair then separate to opposite ends of the cell, and the cell divides between them, giving two new cells with two new nuclei.

10   This is the end of division I of meiosis. At this stage each newly-formed cell contains only one of each pair of homologous chromosomes, and each chromosome consists of two chromatids. As a result of crossing over, a chromosome differs in the material it contains at the end of division I from that at the outset of meiosis.

11   During division II of meiosis the two chromatids of each chromosome separate and move to opposite ends of the cell. The cell divides between these two ends giving two new haploid cells.

12   The movements of the chromosomes during meiosis are brought about by fine fibres made of the protein tubulin. The fibres form a spindle running along the length of the cell, plus kinetochore fibres attached to the chromosomes.

13   The four phases within each division of meiosis are called, respectively: prophase, metaphase, anaphase and telophase.

SAQ 3   Meiosis contains two anaphases: anaphase I and anaphase II, each of which partitions the chromosomal material of the cell into two equal halves. Why, therefore, does meiosis not result in the production of cells with one-quarter of the original diploid number of chromosomes rather than one-half?

SAQ 4   When crossing over occurs, does the exchange of material happen (i) between homologous or non-homologous chromosomes; (ii) between chromatids from the same or from different chromosomes; (iii) during prophase I or prophase II?

SAQ 5   Why does crossing over not happen between the chromatids of homologous chromosomes during division II of meiosis?

4 MEIOSIS AND THE INHERITANCE OF CHARACTERS

This Section brings together the observations made by Mendel (Section 2) and the details of meiosis (discussed in Section 3). Thus, it explains the inheritance of a single pair of contrasting characters in terms of the events that take place within cell nuclei during meiosis.

4.1 A MODEL OF INHERITANCE

The summary at the end of Section 2 lists the features of breeding experiments that any successful theory of inheritance must explain. These are repeated here in abbreviated form in Table 2.

TABLE 2   A summary of the main features of the breeding experiments on maize and peas

1	Existence of pure-breeding strains for a particular character
2	Disappearance of one parental character in the first offspring generation when parents differ in a character for which each is pure-breeding
3	Independence of the outcome of a cross of which parent provides the eggs and which the pollen
4	Reappearance of the vanished character in about one-quarter of the second generation of offspring
5	Departure from an exact 3 : 1 ratio in the phenotypes of the second generation of offspring

How can such features be explained given what is known about DNA, chromosomes and their behaviour during meiosis? DNA is the material of inheritance. It is passed on from parents to offspring through the gametes. What is more, DNA is responsible for the synthesis of non-DNA material. As explained in Unit 19, DNA controls the synthesis of proteins, which in turn control the building-up of the animal's or plant's body. So DNA contributes in a crucial way to the phenotype of the organism.

Consider again the grain colour of maize. This colour is inherited: purple grains of pure-breeding plants develop into plants which themselves have purple grains, and white grains develop into plants that themselves have white grains. Because grain colour is inherited in this way, the DNA of purple-grained maize must be different in some way from the DNA of white-grained maize. The DNA in purple grains must give instructions for the synthesis of a purple pigment, and the DNA in white grains must give instructions that result in white grains. What are these instructions and where do they lie along the DNA? For the moment, let us assume that the colour of a maize grain is determined by a simple instruction: 'You will be purple' or 'You will be white', that this instruction is issued in some way by the DNA, and that there is one particular place along the length of the DNA molecule of one particular chromosome that is responsible for issuing instructions about grain colour (as opposed to, say, height of the plant, shape of the leaves, or some other phenotypic character). This part of the chromosome that issues a particular instruction is called a **gene**. (Figure 21).

This simple assumption that a gene is a chunk of DNA on one particular chromosome and issues instructions for a particular phenotypic character is the starting point of a theory of inheritance. It is the foundation of a genetic model that will be built up gradually during the rest of this Unit. As the Unit proceeds, you should note how the model has to be adapted, and made increasingly sophisticated in order to cope with the increasingly diverse results obtained in breeding experiments.
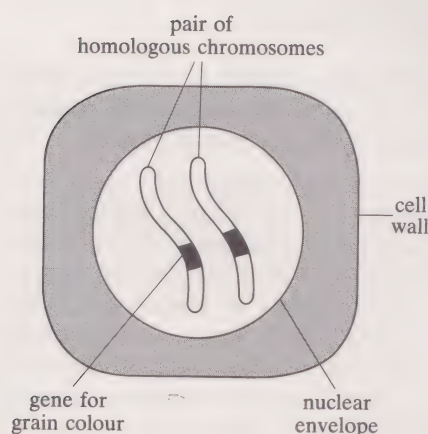

FIGURE 21   The starting point for a genetic model: a gene is a chunk of DNA. For simplicity, only one pair of homologous chromosomes and only one pair of genes is shown.
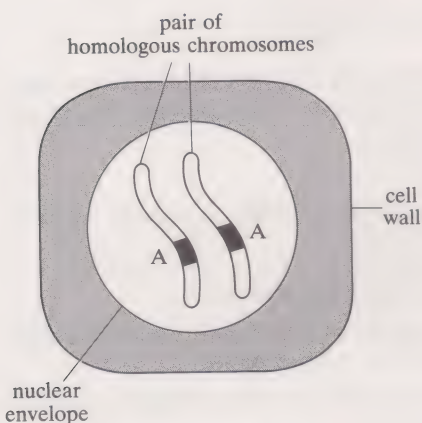
23

LOCUS

GENOME

ALLELES

GENOTYPE

FIGURE 22 A pair of genes for a given character occur at locus A of the respective chromosomes in the homologous pair.

4.2 THE MODEL AND PURE-BREEDING STRAINS

We start by considering the concept of the gene in the light of what we know about chromosome structure. Each chromosome in a diploid cell resembles only one other in that cell, that is, its homologous partner. All of the other chromosomes are different in their shape, size and banding pattern. Consider a gene responsible for issuing instructions for a maize grain to be purple (which, for brevity, can be called a 'gene for purple grains').

If that gene is a chunk of DNA of a particular chromosome, such as band A on the left-hand chromosome in Figure 22, then that gene should exist as the corresponding chunk of DNA of the homologous chromosome. This is band A on the right-hand chromosome in Figure 22. That is, a diploid cell should contain a *pair of genes* for a particular character situated at corresponding locations on the two homologous chromosomes. The technical word for the location of a gene on a chromosome is **locus**.

A single chromosome carries many genes, of the order of 1 000 to 2 000 in a human chromosome, each with its own function, and each with its own specific location, its own locus, on the chromosome. In a pair of homologous chromosomes, there will be two sets of genes strung along their length. Each pair of homologous chromosomes will have its own unique paired strings of genes. The complete collection of genes in a cell is called the **genome**. In summary, the diploid genome consists of two copies of every gene (with the exception of the sex chromosomes—see Figure 6, p. 9), one of each pair at the same locus on each homologous chromosome.

☐ Where do the two copies of a gene in a diploid cell originate?

■ One comes from the mother, the other from the father. Both the mother and the father produce haploid gametes by meiosis, and the diploid condition is restored at fertilization. The zygote then divides repeatedly while developing into the mature organism, and each division produces diploid cells whose genome is identical to that of the zygote.

☐ Why is the outcome of the breeding experiments on maize and peas described in Section 2 the same regardless of which of the parental phenotypes provides the eggs and which the pollen? (item 3 in Table 2)

■ The simplest explanation is that the instructions that a gene issues are the same, irrespective of whether the gene comes from the male or the female parent. For example, the instructions to produce a purple grain will be the same whether the gene is on a chromosome in the ovule, or on the homologous chromosome in the pollen grain.

Consider now what happens when pure-breeding plants are crossed. You may remember from Section 2.1 that when pure-breeding purple-grained plants are crossed with each other, all the offspring are purple-grained.

Figure 23 (opposite) shows what must be happening to the genes for purple grains during the breeding experiment. Each circle represents a cell nucleus. For simplicity, only one pair of homologous chromosomes is shown in the nucleus. Each chromosome of the pair carries a copy of the gene for purple grains. These appear in the Figure as bands labelled *P*. Meiosis produces haploid gametes, and each gamete will have a *P* gene. It does not matter if crossing over occurs during meiosis between the two homologous chromosomes in Figure 23, since this will simply result in the chromatids concerned exchanging one *P* gene for another, as you can see in Figure 24.
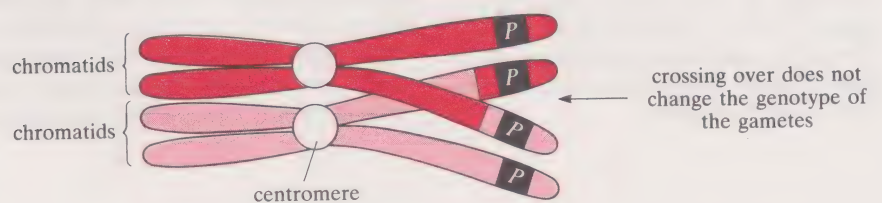
FIGURE 24 Although crossing over results in the exchange of genes between chromosomes, the end result in this example is the same as if no crossing over had occurred, since all four chromatids carry the same gene, *P*, for purple grains.
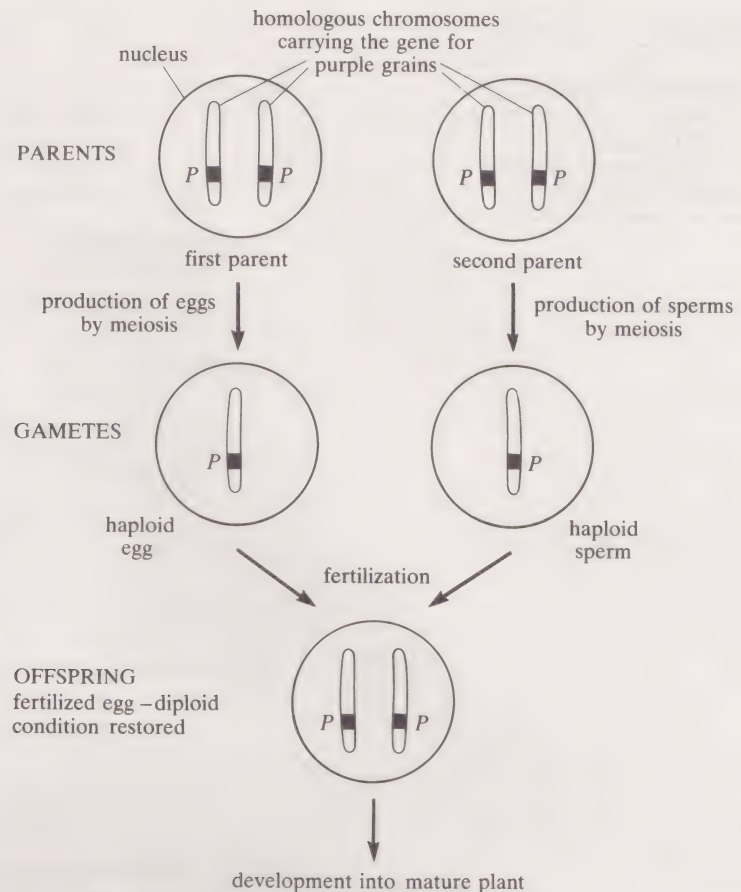
FIGURE 23 The fate of chromosomes and genes during gamete production and crossing of two pure-breeding plants from a purple-grained strain. *P* represents the gene for purple grains.

When fertilization occurs, the diploid condition is restored, and both of the homologous chromosomes will bear the *P* gene, just as in the parental generation. In consequence, their grains will be purple, just like their parents'.
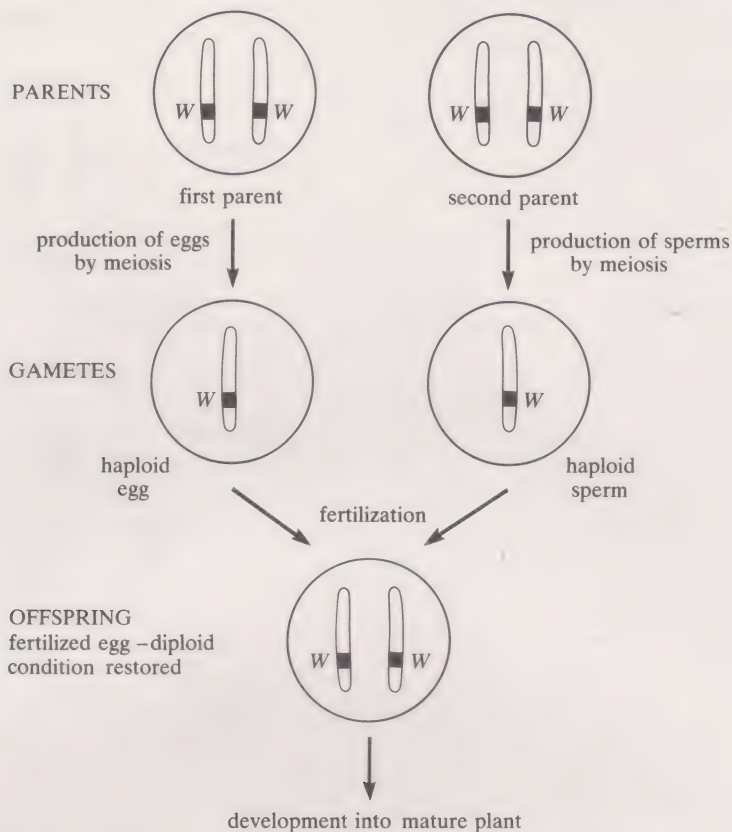
The simple genetic model of pairs of identical genes operating at the same locus on homologous chromosomes therefore successfully explains the phenomenon of pure-breeding, which is item 1 in Table 2.

☐ Repeat the exercise above for white-grained rather than purple-grained plants. Using the letter *W* (for white) rather than *P*, construct a diagram along the lines of Figure 23, modified appropriately.

■ Your diagram should be very nearly identical to Figure 23. This is because the genes controlling grain colour in the offspring are identical to those in the parents. Our version of this diagram is produced overleaf as Figure 25.

If you compare Figures 23 and 25, you can see that the only difference is that they deal with different versions of the gene for grain colour. The different versions of a gene are, technically, called **alleles**. Figure 23 shows the allele for purple grains and Figure 25 the allele for white grains. Notice particularly that the allele for white grains occupies the same locus on the chromosomes as the allele for purple grains. This is part of the definition of an allele. Alleles are different versions of a gene that can occur at the locus occupied by that gene. The alleles that are present in the offspring generation in Figure 23 are the same as those in the parents. To introduce a new term, the parents and offspring have the same **genotype**. The alleles of the offspring in Figure 23 are different from those of the offspring in Figure 25. The two sets of offspring have different genotypes.
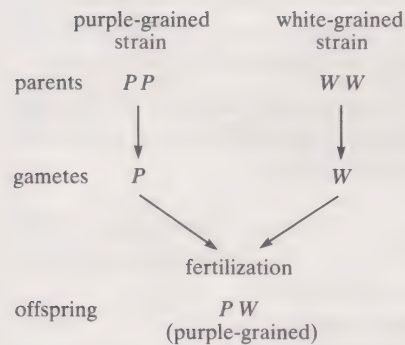
RECESSIVE CHARACTER

DOMINANT CHARACTER

HOMOZYGOUS

HETEROZYGOUS

PARENTS

first parent   second parent

production of eggs   production of sperms
by meiosis   by meiosis

GAMETES

haploid   haploid
egg   sperm

fertilization

OFFSPRING
fertilized egg – diploid
condition restored

development into mature plant

FIGURE 25   The fate of chromosomes and genes during gamete production and crossing of two pure-breeding plants from a white-grained strain. W represents the gene for white grains.

purple-grained   white-grained
strain   strain

parents PP WW

gametes P W

fertilization

offspring PW
(purple-grained)

FIGURE 26   The fate of genes where plants from a pure-breeding purple-grained and a pure-breeding white-grained strain are crossed.

4.3 DOMINANT AND RECESSIVE CHARACTERS

Now consider the situation where plants from the pure-breeding purple-grained strain are crossed with plants from the pure-breeding white-grained strain. Figure 26 depicts what will happen to the genes according to the simple genetic model in which a gene is a chunk of DNA on a chromosome and is responsible for determining an organism's phenotype. The conventions in Figure 26 are similar to those in Figures 23 and 25, but some of the pictorial frills have been left out so that you can concentrate on the genes.

Remember from Section 2.1 that all of the grains in the offspring generation are purple.

☐ Does the model, as depicted in Figure 26, adequately explain the phenotype of the offspring?

■ No. According to the model the genotype of the offspring should be PW, and one might expect the grains with this genotype to manifest the influence of both alleles. Each grain might, for example, be pale purple, or be a mosaic of purple and white patches. One would not expect one of the parental characters to disappear entirely.

Clearly the model will not do in its present form. Perhaps it can be modified, however. Consider what happens if we assume that the two alleles are *not* equal in their phenotypic effect, but that one conceals the effect of the other: the P allele conceals the effect of the W.

☐ Does this modified model account for the phenotypes depicted in Figure 26?

■ Yes. The pure-breeding purple-grained parents have two P alleles. The white-grained parents, with two W alleles, have no P allele to conceal the effect of the W allele, and so are white-grained. The offspring have

both *P* and *W* alleles; the *P* conceals the *W*, and hence the offspring are purple.

The allele whose effect is concealed in the first offspring generation is said to be **recessive**. The other allele, whose effect is shown in the first offspring generation is said to be **dominant**. The terms dominant and recessive are also applied to the *characters* controlled by the alleles.

☐ Which of the garden pea's characters in Table 1 (p. 6) are dominant?

■ Round seeds, yellow seeds, inflated pods, green pods, purple flowers, flowers along the stem and long stems. All of these characters appear in all of the first generation of offspring.

This modification to the genetic model successfully explains item 2 in Table 2 (p. 23); that is, the disappearance of one of the parental characters in the first generation of offspring, and the absence of intermediates between the two parental phenotypes.

Notice some additional features of Figure 26. Both of the alleles in the parent with the *PP* genotype are the same, as are both alleles in the parent with the *WW* genotype. This is not true of the offspring, where the genotype is *PW*; here the two alleles are different. Where the two alleles are the same, the animal or plant is said to be **homozygous** at the locus under consideration. Where they are different, the animal or plant is said to be **heterozygous**. Thus in Figure 26 both parents are homozygous whereas the offspring are heterozygous. Notice also the letters that have been used in Figure 26 to denote purple grains (*P*) and white grains (*W*). This is not the conventional way to denote alleles. The standard convention, which will be used from this point on in the Course, is for the dominant allele to be denoted by a capital letter that readily brings to mind the character in question (usually the initial letter of the character). Hence *P* would denote the allele for purple grains. The recessive allele is denoted by the lower case letter used for the dominant allele. Hence the allele for white grains is denoted by *p* (and not by *W*, as has been the case up till now).

Another convention used in diagrams such as Figure 26 is the use of a multiplication sign to represent a breeding cross. You are familiar (from Section 2) with the use of P, F_1, and F_2 to represent the various generations. With these conventions in mind, Figure 26 can be simplified to the form shown in Figure 27.

P $PP \times pp$

gametes   *P*   *p*

F_1 *Pp*

FIGURE 27   Simplified convention for denoting a breeding cross.

4.4 CROSSING INDIVIDUALS FROM THE FIRST FILIAL GENERATION

Can the modified model of inheritance explain item 4 in Table 2? That is, can it account for the reappearance of the recessive character in the F_2 generation, and for the fact that three times as many individuals in the F_2 generation have the dominant character as have the recessive character?

Consider what happens when the heterozygous F_1 plants produce gametes. Figure 28 (on p. 29) is similar to Figure 10, which you met in Section 3.2 when dealing with meiosis, but the two alleles *P* and *p* are marked on the chromosomes: *P* on the chromosome derived from the mother and *p* on the chromosome from the father. Follow the diagram down through its various stages until you come to stage 6, at the end of the first cell division. Notice that each newly-divided cell carries two copies of the gene for grain colour, one on each chromatid. Notice also that the alleles on the two chromatids from the same chromosome are different, *P* and *p*. This is simply because, as the diagram shows, the parts of the chromatids carrying *P* and *p* crossed over at stage 3. If crossing over had *not* occurred, the alleles on the two chromatids from the same chromosome would have been the same: *PP* on the maternal chromosome and *pp* on the paternal one. Regardless of whether or not crossing over has occurred, the effect of division II of

27

meiosis is the same (stages 7–9). It splits the two chromatids apart (stage 8) so that each gamete receives only one copy of the gene for grain colour. Meiosis therefore results in the separation (or *segregation*, as it is more usually called) of alleles that were formerly together in the same cell.

☐ What proportion of the gametes so produced carry the *P* allele and what proportion carry *p*?

■ Half of them carry the *P* allele and half carry *p*.

You can see this by looking at the bottom left of Figure 28. The two cells at stage 8 divide to give a total of four gametes, two containing the *P* allele and two containing *p*.

Let us now imagine what happens during the production of the maize cob A in your Kit. (Remember that this is an F_2 cob, produced as a result of the transfer of F_1 pollen to F_1 eggs.) The young cob is made up of a dense cluster of many flowers, each of which produces a single egg. Once a cloud of pollen has landed on the cluster of flowers in the flower head, hundreds or thousands of male gametes race down their pollen tubes towards each ovule. In the case of the F_1 plants, half of these eggs will carry the *P* allele and half will carry *p*. Exactly the same applies to the male gametes of the F_1 plants. Suppose that the few male gametes that succeed in fertilizing the eggs do so irrespective of whether they carry the *P* or the *p* allele, and that they are equally likely to fertilize an egg irrespective of whether the egg carries *P* or *p*. Suppose, that is, that fertilization occurs *randomly* with respect to the *P* and *p* alleles. (This is as an additional assumption that has to be incorporated into the genetic model that we are developing.) What will be the genotypes of the fertilized eggs, what will be their phenotypes, and in what proportion will they occur?

These questions can most easily be answered by first drawing up a blank Table like Table 3.

Along the top of the Table you can see the two types of female gametes; down the left-hand side you can see the two types of male gametes. The Table can be used to show the outcome of fertilization between the various kinds of gametes. Thus, box 1 records the outcome of fertilization between a *P* ovule and a *P* sperm.

Table 3   For ITQ 3.

genotypes of
female gametes

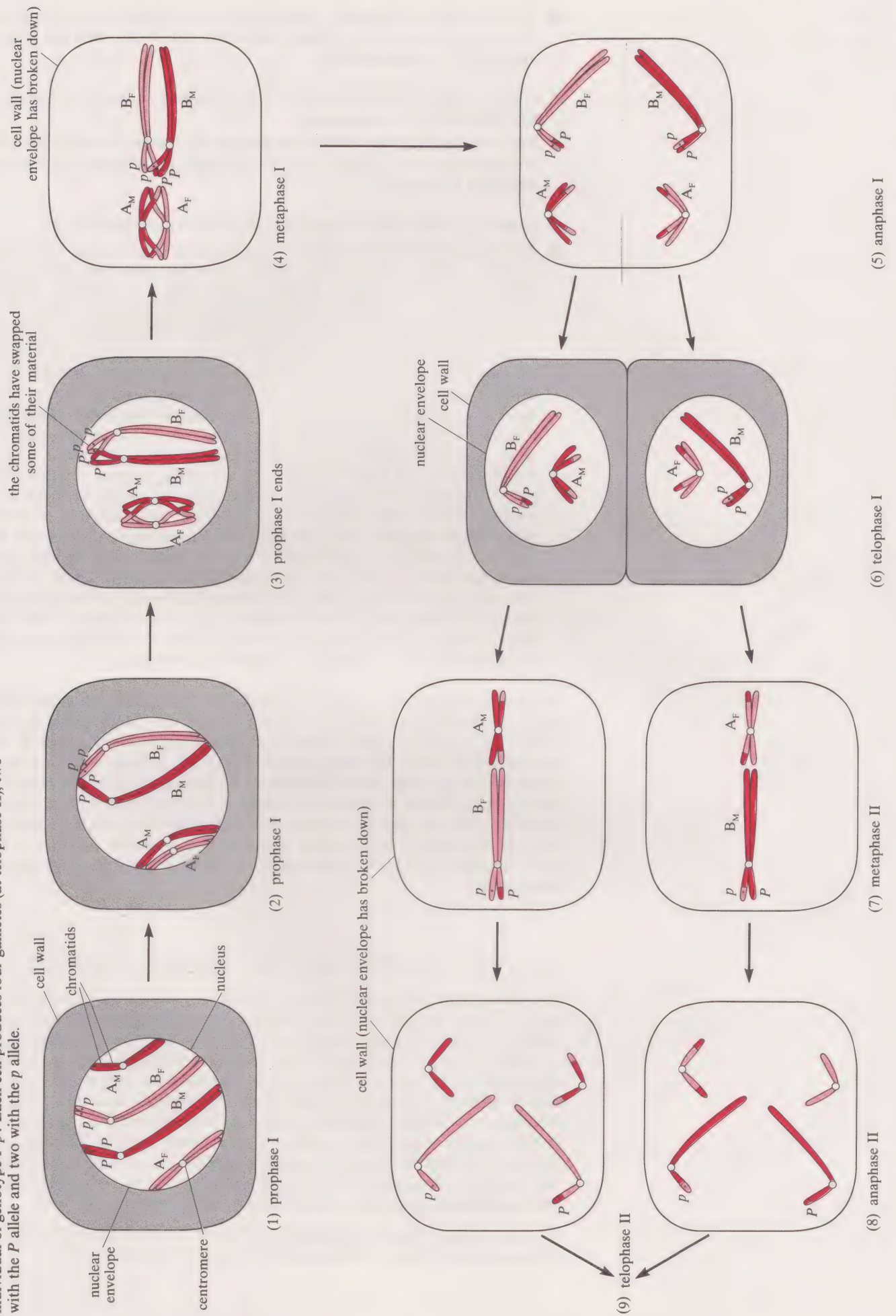|	*P*	*p*
P	PP Purple ₁	Pr Purple ₂
P	p P Purple ₃	Pr white ₄

genotypes of male gametes

ITQ 3   Complete Table 3 to show the genotype and the phenotype that would result from each of the four combinations of gametes.

Note: You should ensure that you have filled in the information correctly before proceeding. This instruction applies also to subsequent ITQs in this Unit.

☐ Does the genetic model explain the reappearance in the F_2 generation of the recessive character?

FIGURE 28 What happens to alleles *P* and *p* during meiosis in a heterozygous individual of genotype *P p*? Each cell produces four gametes (at telophase II), two with the *P* allele and two with the *p* allele.

(1) prophase I

(2) prophase I

(3) prophase I ends

the chromatids have swapped some of their material

(4) metaphase I

(5) anaphase I

(6) telophase I

(7) metaphase II

(8) anaphase II

(9) telophase II

cell wall (nuclear envelope has broken down)

nuclear envelope

cell wall

cell wall (nuclear envelope has broken down)

nuclear envelope

cell wall

chromatids

nucleus

centromere

■ Yes. The recessive character reappears in those individuals in which the dominant allele is totally absent, that is, in individuals that are homozygous for the recessive allele.

☐ Does the genetic model explain the 3 : 1 ratio of dominant to recessive characters in the F_2 generation.

■ Yes, provided that the model incorporates the assumption that the possession of a P or a p allele does not influence which sperm succeeds in fertilizing which egg.

☐ Does the genetic model explain why the ratio is not *exactly* 3 : 1?

■ Yes. Suppose that exactly half of the eggs on a maize flower head contained P and the other half contained p. Imagine also that of the thousands or perhaps millions of pollen grains that are artifically dabbed onto the stigmas of the flowers in the breeding experiment, exactly half contain P and the other half contain p. Which of these millions of pollen grains produces the few hundred gametes that win the race to fertilize the eggs is totally independent of whether those gametes contain P or p. Purely by chance, the proportion of P-containing gametes that succeed in this way may be rather higher than one-half for one flower head. The cob would then develop rather more than the 3 : 1 ratio of purple to white grains. On another flower head the opposite might occur, with a disproportionately higher number of p-containing sperms fertilizing the ovules. Similarly, the proportion of P-containing ovules fertilized may be more (or less) than one-half. If a sufficiently large number of flower heads are investigated, then the ratio will be very close to 3 : 1, but for any one flower head and hence for any one cob, it may deviate quite markedly from 3 : 1. The situation is similar, in principle, to the tossing of a coin. If you were to toss a coin one million times, you would expect the ratio of heads to tails to be close to 1 : 1. If you tossed it only ten times, however, you would not be very surprised to find quite a marked departure from a 1 : 1 ratio (i.e. five heads and five tails).

The genetic model that we have developed therefore accounts successfully for all of the items in Table 2. The model states: that a character is controlled by a pair of genes, one on each homologous chromosome at the corresponding locus; that the genes exist as two alleles, with one allele dominant to the other; that the genotype of the gamete does not influence its success or failure to achieve fertilization. Although this model is more elaborate than the 'gene-is-a-chunk-of-DNA' model with which we started, it is still very simple. Nevertheless it is also very powerful, as you can see from the impressive list of phenomena for which it provides an adequate explanation.

4.5 THE IMPORTANCE OF MENDEL'S WORK

The genetic model described in the last Section could not have been developed without the work of Gregor Mendel. Mendel's experiments with peas, some of whose results are summarized in Table 1, were the beginning of the systematic study of inheritance. When Mendel published his results in 1865 neither he nor anyone else knew about chromosomes, and so he was not able to explain his results in terms of known physical structures. Despite this handicap, Mendel was able to postulate from his breeding experiments: (a) the existence of genes (or 'particles of inheritance', as he called them); (b) that genes are present in pairs in parents; (c) that the genes separate during the formation of gametes, so that each gamete contains only one of each pair; and (d) that genes have alternative dominant and recessive forms.

This was a phenomenal achievement on Mendel's part. William Bateson, a leading geneticist from a later generation, wrote of Mendel in 1913:

His success was due to the clearness with which he thought out the problem ... he surmised that the failure [of earlier scientists] to reach definite and consistent conclusions was due to a want of precise and continued analysis. In order to obtain a clear result he saw that it was absolutely necessary to start with pure-breeding homogeneous materials, to consider each character separately and on no account to confuse different generations together. Lastly he realised that the progeny from distinct individuals must be separately recorded. All these ideas were entirely new in his day.

(Bateson, 1913)

The importance of Mendel's achievement was not realized immediately after he published his results in 1865. In fact, it was not until 1900, 16 years after his death, that his work began to be understood. Just why it took 35 years for the scientific community to begin to appreciate Mendel's achievments has never been explained satisfactorily. One reason may be, although this has been disputed, that he published his work in an obscure journal in the Moravian (now Czechoslovakian) town of Brno, and hence his work was not widely known. Another reason may have been that scientists of the day were simply not interested in or receptive to his ideas.

Whatever the reason, the neglect of Mendel's work delayed advances in the understanding of the mechanism of natural selection. You may remember from Unit 19 that the theory of evolution by natural selection was first put forward by Darwin and Wallace in 1858—only seven years before the publication of Mendel's work. Yet although Darwin, Wallace, and those who followed their ideas, realized that the inheritance of characters was central to their theory, they had no clear idea about the *mechanism* of inheritance. Indeed, Darwin believed that the phenotypic variation that was important for natural selection was inherited in a blending fashion.That is, he thought that if two parents differed in a phenotypic character that significantly affected their fitness, and if they were cross-bred, then the offspring would show a phenotypic character that was intermediate between that of both parents. This idea presents considerable problems to the theory of natural selection, because if a new and favourable character were to arise through mutation and if the organism carrying the character were to breed with non-mutant individuals (these being in the great majority in the population), the new, favourable, character would appear in less pronounced form in the offspring. If these offspring in turn bred, *their* offspring would display the character still less distinctly, and after several generations the mutant character would have become so diluted as to become virtually unnoticeable. The variation so essential to natural selection would thereby have vanished.

In order to counteract this supposed continuous dilution of mutant characters, Darwin was led to postulate that new characters appeared in a population very frequently, at a far higher rate than (as we now know) mutations occur. These difficulties disappear as soon as one accepts that inheritance is not blending. In other words, if a character that is heritable appears in a population, then it will not be diluted from one generation to the next. Variation will not decline as the generations pass, and natural selection will be able to continue to operate on this variation.

SUMMARY OF SECTION 4

1 This Section develops a genetic model that can account for some of the results of breeding experiments involving maize and garden peas. The five features to be accounted for are those in Table 2 (p. 23).

2 The first of these features is the existence of pure-breeding strains for a particular character.

The model accounts for this by postulating:

(i) that a particular phenotypic character is determined by a pair of alleles at a particular locus on a pair of homologous chromosomes; and

(ii) that, in a pure-breeding strain, these two alleles are identical and remain so when individuals of that strain breed with each other.

3   The second feature is that when individuals that differ in a character for which each is pure-breeding are crossed, one parental character is entirely absent from the F_1 generation.

The model accounts for this by postulating that the two contrasting characters are determined by two different alleles, one being dominant, the other recessive. In the heterozygous condition (that is, where there is one dominant allele and one recessive allele at a particular locus), the resulting phenotype is always that of the dominant allele. Thus, only the character shown by the parent homozygous for the dominant allele will appear in the F_1 offspring.

4   The third feature is that the outcome of a cross between parents differing in a particular character is independent of which (of a pair of contrasting characters) is possessed by the mother and which by the father.

The model accounts for this by postulating that one allele carries the instructions for one character, and that these instructions are not affected by whether that allele was present in the male gamete or in the female gamete before fertilization.

5   The fourth feature is that the recessive character (the one that vanishes at F_1) is shown in about one-quarter of the F_2 offspring, the other three-quarters or so being individuals with the dominant character of the contrasting pair.

The fifth feature is that the F_2 generation does not contain an exact 3 : 1 ratio of dominant to recessive phenotypes.

These last two features are accounted for in the model as follows.

In individuals heterozygous at a particular locus, half the gametes will contain one allele and half will contain the other allele. Where both parents are heterozygous, assuming fertilization is a random process that is not influenced by the genes carried by the gametes, about one-quarter of the offspring will be homozygous for the dominant allele, about half will be heterozygous and about one-quarter will be homozygous for the recessive allele. This explains why about three-quarters of the F_2 offspring show the dominant character and about one-quarter show the recessive character. The random nature of fertilization accounts for small deviations in the 3 : 1 ratio.
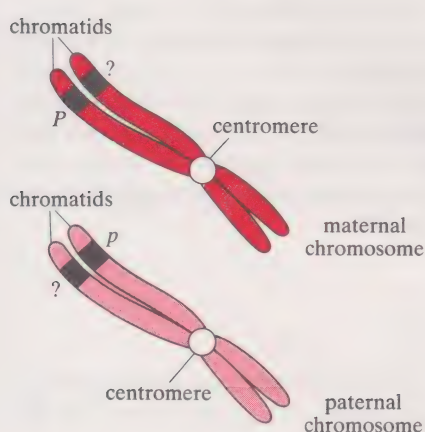

FIGURE 29   The chromatids of one pair of homologous chromosomes at the start of meiosis.

SAQ 6   A breeding experiment was carried out in which plants from a pure-breeding purple-grained strain of maize with genotype PP were crossed with heterozygous plants with genotype Pp. What genotypes and phenotypes would occur in the offspring arising from this experiment, and in what ratios would they occur?

SAQ 7   A breeding experiment was carried out in which plants from a pure-breeding white-grained strain of maize were crossed with heterozygous plants of genotype Pp. What genotypes and phenotypes would occur in the offspring arising from this experiment, and in what ratio would they occur?

SAQ 8   Figure 29 shows the chromatids of one pair of homologous chromosomes in a heterozygous Pp maize plant at the start of prophase I of meiosis. Complete the Figure to show which alleles occupy the unlabelled loci (loci is the plural of locus). Is there more than one correct answer to this question? Check your answer before going to SAQ 9.

SAQ 9   Could the arrangement of alleles on the chromatids be different at the end of prophase I from the one shown in the answer to SAQ 8? If so, how could it have arisen?

5 THE INHERITANCE OF MORE THAN ONE CHARACTER

This Section examines slightly more complicated examples of inheritance and shows how they can be explained in terms of the events that take place within cell nuclei during meiosis.

5.1 INDEPENDENT ASSORTMENT

Mendel first investigated the inheritance of single pairs of contrasting characters, such as round versus wrinkled seeds (Section 2.2); later he studied the simultaneous inheritance of more than one pair of contrasting characters. His results revealed some more fundamentally important features of inheritance. Once again, these features were not fully understood until the discovery, many years later, of the behaviour of chromosomes during meiosis. In this Section we are going to reverse history and ask you to predict, from your knowledge of meiosis, the outcome of breeding experiments involving two pairs of contrasting characters. You will be able to check your predictions by investigating maize cob B which is in Tray C of the Experiment Kit.

If you look at maize cob B you will see that not only are some grains purple and others white, but that some are smooth and others wrinkled. Thus there are two pairs of contrasting characters. The two alleles for grain colour (purple and white), as before, are denoted by P and p, respectively; those for smooth and wrinkled by S and s, respectively. The purple character is dominant to the white, and the smooth character dominant to the wrinkled. Maize cob B is from the F_2 generation. In the parental generation, pure-breeding plants from a strain that always produced purple, smooth grains were crossed with pure-breeding plants from a strain that always produced white, wrinkled ones.

☐ Describe the different kinds of grains on maize cob B. Which are the most common and which the least?

■ There are four kinds of grains: purple, smooth ones, which are the most common; purple, wrinkled ones and white, smooth ones, which are much less common; and white, wrinkled ones, which are rather rare.

Bearing this general picture in mind, consider what will happen during meiosis to the genes for two pairs of contrasting characters. First, we have to make an important decision: should we consider what happens to two pairs of genes that happen to lie on the *same* homologous pair of chromosomes, or should we consider those that lie on *different* pairs of chromosomes? The two alternatives are shown in Figures 30 and 31.
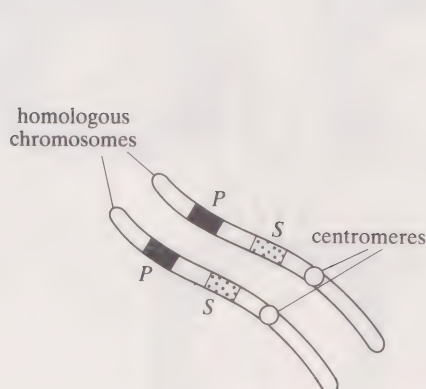

FIGURE 30 The genes for two pairs of contrasting characters (grain colour and coat texture) shown situated on the *same* pair of homologous chromosomes.
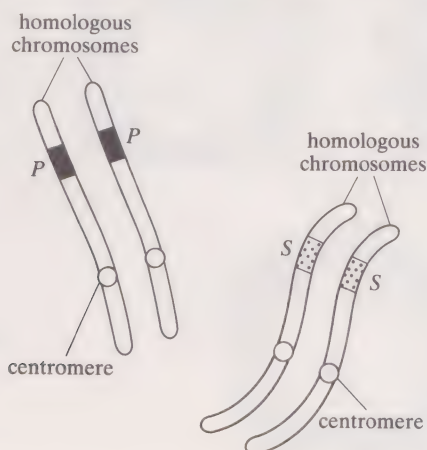

FIGURE 31 The genes for two pairs of contrasting characters (grain colour and coat texture) shown situated on *different* pairs of homologous chromosomes.

purple, smooth grains

white, wrinkled grains

prophase I

anaphase I

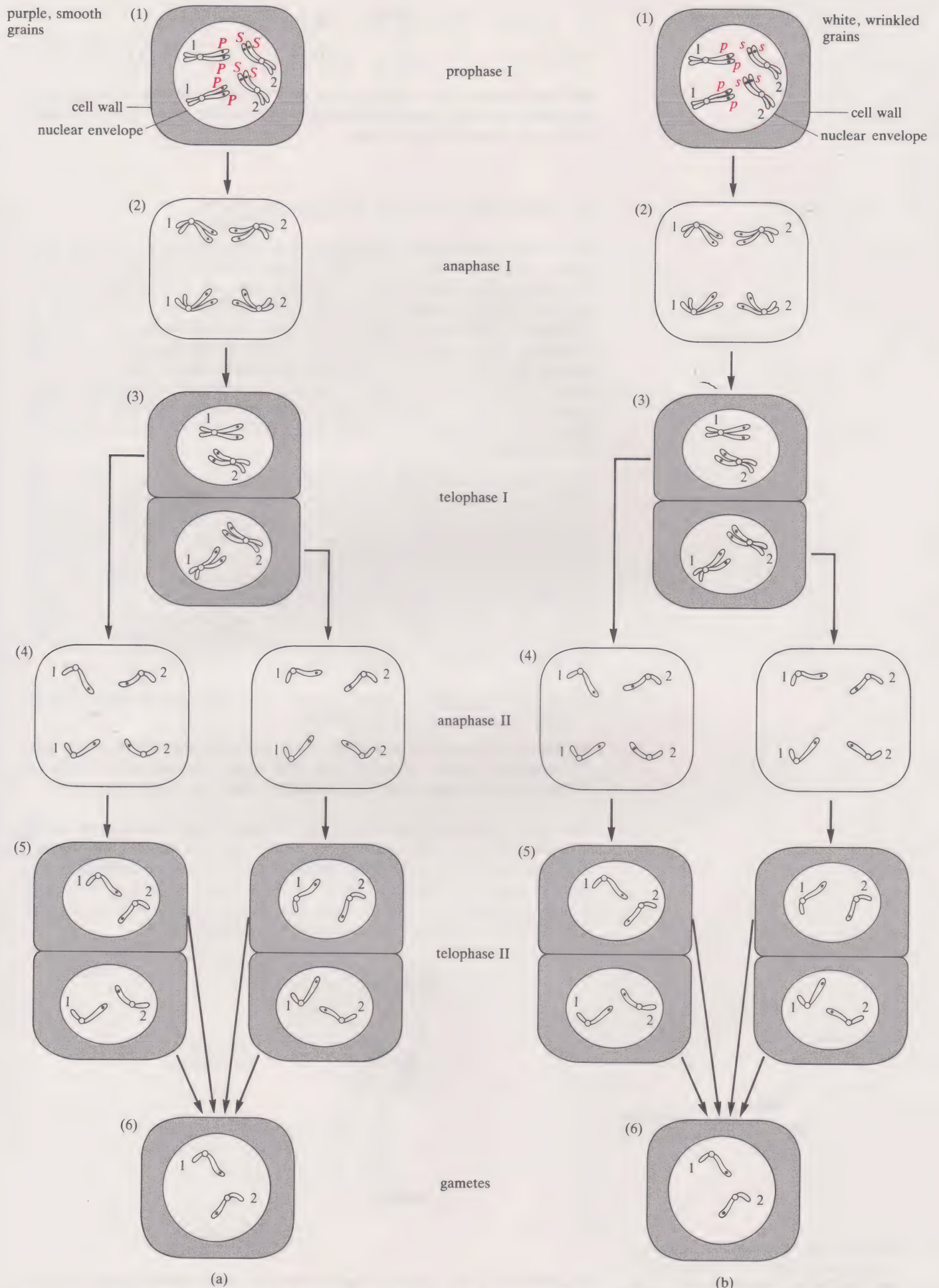telophase I

anaphase II

telophase II

gametes

(a)

(b)

FIGURE 32 The fate of two pairs of genes on different pairs of homologous chromosomes during meiosis. (a) Meiosis in a pure-breeding strain of maize with purple, smooth grains; (b) meiosis in a pure-breeding strain of maize with white, wrinkled grains.
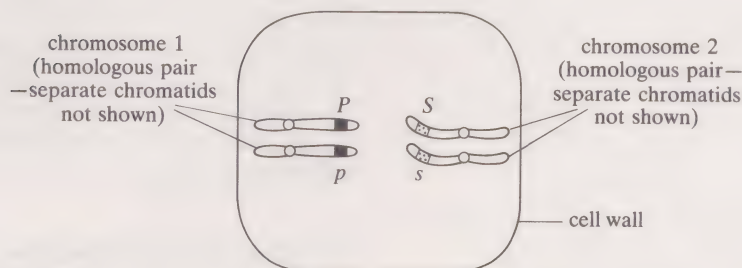
For the moment we shall concentrate on the second of these two possibilities; pairs of genes on different pairs of homologous chromosomes. Suppose, therefore, that the genes for grain colour are on one pair of chromosomes (which can be called chromosome 1) and the genes for coat texture (smooth or wrinkled) are on another pair (chromosome 2). Figure 32 (stage 1) shows these two pairs of chromosomes in a breeding experiment between two parent plants, one of them pure-breeding with purple, smooth grains, the other pure-breeding with white, wrinkled grains. Stages 2–6 of Figure 32 show what happens to the two pairs of chromosomes during meiosis. Look at this diagram carefully; then, using the standard letters P, p, S and s for the alleles, do the following ITQ.

ITQ 4   Add to Figure 32 the letters for the alleles at each stage during meiosis in the parent with purple, smooth grains (a) and in the parent with white, wrinkled grains (b) and so show the alleles that each type of gamete possesses at the end of meiosis. Then refer to the ITQ 4 answer on p. 53.

You can see from your completed Figure 32 that the parent from the purple, smooth-grained strain will produce gametes in which chromosome 1 always carries the P allele, and chromosome 2 always carries the S allele. Even if there is crossing over during meiosis, the outcome will be the same. Similarly, the parent from the white, wrinkled-grain strain will produce gametes in which chromosome 1 always carries the p allele, and chromosome 2 always carries the s allele.

☐ What will be the genotype of the F_1 generation resulting from a cross between these two parents?

■ The genotype will be $P p S s$. Fertilization restores the diploid condition, with each chromosome bearing a different allele from its homologous partner.

☐ What will the grains of the F_1 generation be like, and how will they compare with those of the parental generation?

■ All of the grains will be purple and smooth. They will therefore resemble the grains of the parent carrying the two dominant alleles.

Now consider what happens when the plants that develop from these F_1 grains are crossed with each other. Figure 33 shows the two pairs of homologous chromosomes in these parents. Before one can work out what happens to the genes on these chromosomes during meiosis, it is necessary to make one very important assumption about the way that chromosomes 1 and 2 line up around the equator of the cell at metaphase I. Figure 33 is drawn in such a way that the chromosomes carrying genes P and S both lie above the equator, and the chromosomes carrying genes p and s both lie below it. However, there are other perfectly valid ways round we could have drawn the Figure, and we need to explore those.

chromosome 1
(homologous pair
—separate chromatids
not shown)

P S

p s

chromosome 2
(homologous pair—
separate chromatids
not shown)

cell wall

FIGURE 33   Two pairs of genes on different pairs of homologous chromosomes at metaphase I of meiosis. The plant is heterozygous at both loci.

INDEPENDENT ASSORTMENT

chromosome 1
(homologous pair)   cell wall

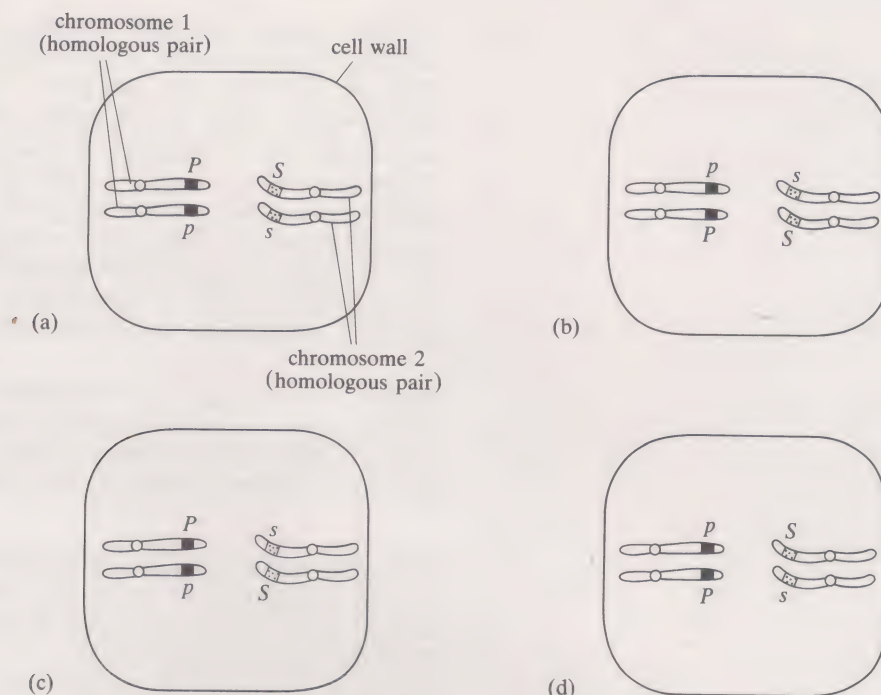chromosome 2
(homologous pair)

Figure 34   The four possible
arrangements of two pairs of
homologous chromosomes at
metaphase I of meiosis.

In fact there are four possible arrangements and these are shown in Figures 34a–d. Figure 34a is, as you can see, identical to Figure 33 (the latter is one of the four possibilities) and 34b is an upside down version of 34a—with the chromosomes carrying genes P and S lying below the equator and the chromosomes carrying p and s above it. Figure 34c shows P and s to be above the equator, and p and S below it, and finally 34d shows p and S above the equator and P and s below it. Which of these four options occurs?

The answer is: All four occur in more or less equal proportions. This is because the position that any pair of chromosomes takes up along the equator is entirely independent of the position of any other pair. As a result, each of the four options in Figure 34 is equally likely to occur. The two chromosomes in pair 1 are as likely to lie with P above the equator and p below, as the other way round. Similarly the chromosomes of pair 2 are as likely to lie with S above and s below, as vice versa. The technical term for pairs of chromosomes sorting themselves out independently in this way is **independent assortment**.

The independent assortment of chromosomes has very important implications for genes borne on those chromosomes, and hence for the inheritance of the characters affected by those genes.

ITQ 5   Look at Figure 35 and consider the different alleles for grain colour and coat texture in stages 2 to 4 of this Figure. As you can see, there are four different options ((a), (b), (c) and (d)) at each stage—each one corresponding to one of the four possible arrangements of chromosomes 1 and 2 around the equator of the spindle. Fill in the letters for the different alleles in options (b) to (d) of Figure 35. Option (a) has been completed for you. Check that you have completed Figure 35 correctly and then answer the following question.

☐ What will be the genotypes of the gametes produced from arrangements (a), (b), (c) and (d), respectively?

■ The gametes produced from arrangement (a) will be of two types, $P S$ and $p s$, in equal numbers. The same is true for the gametes produced from arrangement (b). The gametes produced from arrangement (c) will be of two types, $P s$ and $p S$, in equal numbers. The same is true for the gametes produced from arrangement (d).
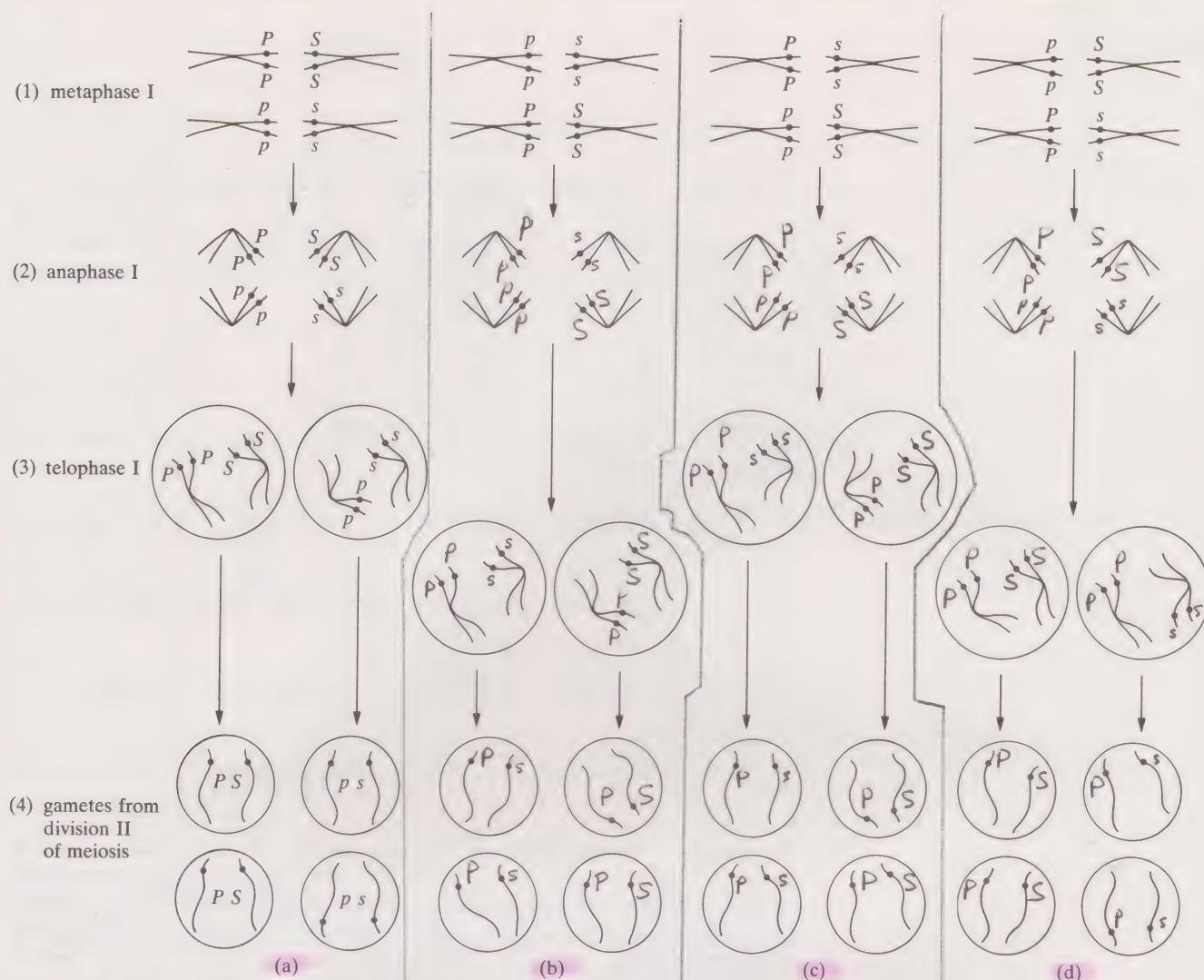
FIGURE 35 **The genotypes of gametes** resulting from each of the four different arrangements of chromosomes at metaphase I of meiosis. (For ITQ 5)

(1) metaphase I

(2) anaphase I

(3) telophase I

(4) gametes from division II of meiosis

(a)   (b)   (c)   (d)

Altogether, therefore, meiosis will have produced four different genotypes in the gametes from the F_1 parents, namely pS, Ps, PS and ps, and these genotypes will be present in equal numbers.

Consider now what happens when the F_1 gametes fuse to produce the F_2 generation. Remember that, according to the genetic model we have developed, the chances of any one sperm fertilizing any one egg is independent of their genotype. This means that each of the four genotypes carried by the pollen has an equal chance of combining with each of the four genotypes carried by the eggs. What are the consequences of this? We can work this out by using the kind of Table shown in Table 4 overleaf.

Each box in the Table is used to write in the genotype of the fertilized egg (zygote) resulting from the fusion of the gametes with the genotypes indicated at the beginning of the row and column in which the box lies. Thus the genotype entered in box 7 results when a sperm with genotype Ps fertilizes an egg with genotype pS, and so on. Remembering that a recessive character does not appear in the grain unless *two* recessive alleles for that character are present, answer the following ITQ.

ITQ 6 Enter in Table 4 the genotypes of the F_2 generation arising from these combinations. Also write in the phenotypes (grain colour and coat texture) corresponding to these genotypes. (Don't forget to check your answer.)

TABLE 4   For ITQ 6

genotypes of
female gametes

	P S	P s	p S	p s
P S	PSPS PPSS *1*	PSPs PPSs *2*	PSpS PpSS *3*	PSps PpSs *4*
P s	Ps PPsS *5*	Ps PPss *6*	PpsS *7*	PPss *8*
p S	pPSS *9*	pPSs *10*	ppSS *11*	ppSs *12*
p s	pPsS *13*	pPss *14*	ppsS *15*	ppss *16*

genotypes
of male
gametes

From your completed Table 4, you can see the following. Each of the 16 combinations is as likely to occur as any other. Also, because the recessive *character* does not appear unless two recessive *alleles* for that character are present in the zygote, the phenotype corresponding to each genotype can be worked out fairly easily. Counting up the phenotypes, you can see that in nine boxes the grains will be purple and smooth; in three boxes the grains will be purple and wrinkled; in three boxes the grains will be white and smooth; and in just one box will the grains be white and wrinkled. In summary, the ratio of the four phenotypes will be 9 : 3 : 3 : 1. Figure 36 summarizes the whole breeding experiment from parental to F_2 generation. The model that we have developed, which now includes the assumption of independent assortment of chromosomes, leads to a precise prediction about the ratios of phenotypes in the F_1 and F_2 generations of any breeding experiment involving two sets of contrasting characters borne on different chromosomes. You can see whether the prediction is correct, and so test the model by counting the number of grains of each of the four phenotypes in your maize cob B.

parental generation	P P S S (purple, smooth)	p p s s (white, wrinkled)
gametes	P S	p s
F_1	P p S s (purple, smooth)	× P p S s (purple, smooth)
gametes	P S P s p S p s	
F_2	purple, smooth : purple, wrinkled : white, smooth : white, wrinkled	
	9 3 3 1	
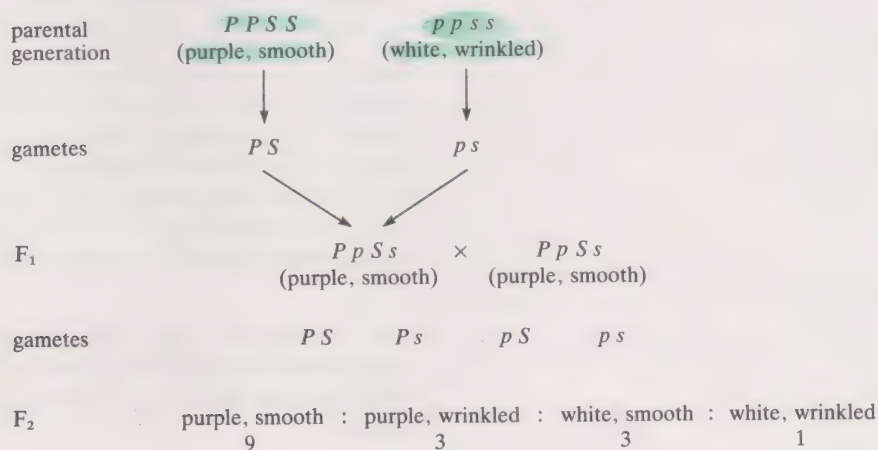
FIGURE 36   Summary of breeding experiment involving two pairs of contrasting characters borne on different chromosomes.

EXPERIMENT 2 GRAIN COLOUR AND COAT TEXTURE IN MAIZE

TIME

This experiment takes about 30 minutes.

NON-KIT ITEM

Felt-tip pen or sticky paper

KIT ITEM

Tray C
Cob B

METHOD

Using the same method as you used in Experiment 1, count the grains in cob B. Enter your results in Table 5 below.

TABLE 5   Number of grains of four different phenotypes in cob B

purple, smooth	purple, wrinkled	white, smooth	white, wrinkled	total

ITQ 7   What ratio of the four phenotypes do you obtain with your cob B? Compare your results with the range of results for similar cobs in Table 11 in the ITQ answer.

Notice that, as with the inheritance of a single pair of contrasting characters (Table 8, p. 53), the results vary from one cob to another, but that the ratio of the four phenotypes is roughly $9:3:3:1$. For example in the cob number 5 in Table 11 there are $307 \div 33 \approx 9.30$ times as many purple, smooth as white, wrinkled grains; $101 \div 33 \approx 3.06$ times as many purple, wrinkled as white, wrinkled grains; and $92 \div 33 \approx 2.79$ times as many white, smooth as white, wrinkled. If you were to examine hundreds or thousands of maize cobs and keep a running total of the four different phenotypes, you would find that their ratio approached more and more closely to $9:3:3:1$, giving one confidence in the genetic model that predicted this ratio.

☐ How do you explain the fact that the ratio is not exactly $9:3:3:1$ in any one cob?

■ There are two reasons.
1   The independent assortment of chromosomes during prophase I of meiosis produces equal numbers of the four types of gametes, *on average*. When millions of gametes are examined, the four types of gametes are likely to be formed in very nearly equal numbers. But if you look at just a few gametes, then, purely by chance, there may be a predominance of those in which, say, the chromosome with the P gene and the chromosome with the S gene lined up together on the same side of the spindle's equator at metaphase I, and rather few in which they lined up on opposite sides. This would give a relative excess of PS and ps gametes, and a relative shortage of Ps and pS. If you look at Table 6 (overleaf) you can see that this will lead to a relative shortage of F_2 individuals with the genotypes shown in the four shaded squares in the middle.

2   Even if the gametes with the four different genotypes were produced in exactly equal numbers, one or more genotypes might, purely by chance, be involved in more fertilizations than others. This is exactly the same phenomenon as you saw in the inheritance of a single pair of contrasting characters.

For both of these reasons, therefore, the actual ratio of the four phenotypes in the F_2 generation is likely to deviate some way from $9:3:3:1$.

genotypes of female gametes

	PS	Ps	pS	ps
PS	$PPSS$	$PPSs$	$PpSS$	$PpSs$
Ps	$PPsS$	$PPss$	$PpsS$	$Ppss$
pS	$pPSS$	$pPSs$	$ppSS$	$ppSs$
ps	$pPsS$	$pPss$	$ppsS$	$ppss$

genotypes of male gametes

TABLE 6 Chance may lead to a relative shortage of gametes with some genotypes (Ps and pS) and hence a relative shortage of offspring with some genotypes (those in the shaded area)

Before moving on, there is one further interesting point about the results of the breeding experiment involving grain colour and coat texture. Look again at Figure 36.

□ What is the ratio of purple to white grains in the F_2 generation? Also, what is the ratio of smooth to wrinkled grains? How do you explain these ratios?

■ From Figure 36 you can see that the ratio of purple to white grains is 12 : 4, or, more simply, 3 : 1. The ratio of smooth to wrinkled grains is also 3 : 1.

You should immediately recognize this 3 : 1 ratio as the one you met in Section 4.4 when looking at the inheritance of a *single* pair of contrasting characters. Remember from Section 4.4 that if F_1 plants that are heterozygous for the character in question are crossed, then the two contrasting characters will be present in the F_2 generation in the ratio of 3 : 1. Indeed, since the very character that was discussed in Section 4.4 *was* grain colour, and since you saw there that the ratio of purple to white grains in the F_2 generation was 3 : 1, you would actually expect to find the same ratio of purple to white grains, regardless of how many other pairs of contrasting characters you were looking at. Just the same argument applies to the ratio of smooth to wrinkled grains.

5.2 LINKAGE

As a final step in the development of the genetic model, consider a breeding experiment that is identical in all its main features to the one described in Section 5.1, but with one important exception. This is that the two sets of contrasting characters, instead of being carried on *different* pairs of homologous chromosomes, are carried on the *same* pair (as in Figure 30).

Instead of using maize to illustrate this situation, it is convenient to choose a favourite animal in genetic research, the fruit-fly *Drosophila*. This is a small, harmless fly, often found round soft, fermenting fruit. One species of fruit-fly *Drosophila melanogaster* (Plate 12) has a large number of pheno-

typic forms. For example, although the wings are normally slightly longer than the body there is one strain, called 'vestigial wing' in which the wings are reduced to stumps. The eyes are normally bright red, but there is a strain, called 'cinnabar eye', in which they are brownish red . Whether a fruit-fly has normal or vestigial wings depends upon which alleles it has at a particular locus. If the dominant and recessive alleles are called W and w respectively, then a fly that is homozygous for the w allele, ww, has vestigial wings, and a fly that is homozygous for the W allele, WW, or one that is heterozygous, Ww, has normal wings. Similarly, if the dominant and recessive alleles for eye colour are called E and e, respectively, then a fly that is homozygous for the e allele, ee, has cinnabar eyes, and a fly that is homozygous for the E allele, EE, or one that is heterozygous, Ee, has normal eyes.

Consider what happens in the following breeding experiment. At the start, there are two, pure-breeding strains of fruit-fly; one strain has normal wings and cinnabar eyes, the other has vestigial wings and normal eyes. Figure 37 shows the genotypes of these two strains, and the genotype of the F_1 offspring that result from crossing them.
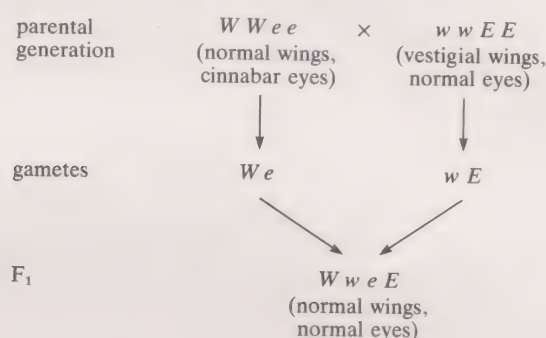
FIGURE 37   The F_1 genotype resulting from a cross between two pure-breeding strains of the fruit-fly *Drosophila melanogaster*, one with normal wings and cinnabar eyes, the other with vestigial wings and normal eyes.

Notice that both parents are homozygous for both genes. If they were not, they would not be pure-breeding. All the gametes from one parent are identical, as are all the gametes from the other parent. As a result, the F_1 generation has only one genotype, in which all flies are heterozygous for both genes. The dominant character appears for both genes, hence the F_1 flies all have normal wings and normal eyes.

Now consider what happens when the F_1 flies are crossed, but this time *not with each other*. Instead, the female F_1 flies are crossed with males that are recessive for both characters. These males therefore have vestigial wings and cinnabar eyes. Figures 38a and b lead you to investigate this cross on the basis of two different assumptions, and so predict two different outcomes.

ITQ 8   Complete Figures 38a and b overleaf. Figure 38a leads you to predict the relative frequency of the different phenotypes that would arise from this procedure if the genes for wing length and eye colour were on *different* chromosomes. Figure 38b leads you to predict their frequency if the genes were to lie on the *same* chromosome. For Figure 38b, assume that the dominant allele W lies on the same chromosome as the recessive allele e, and that the recessive allele w lies on the same chromosome as the dominant allele E.
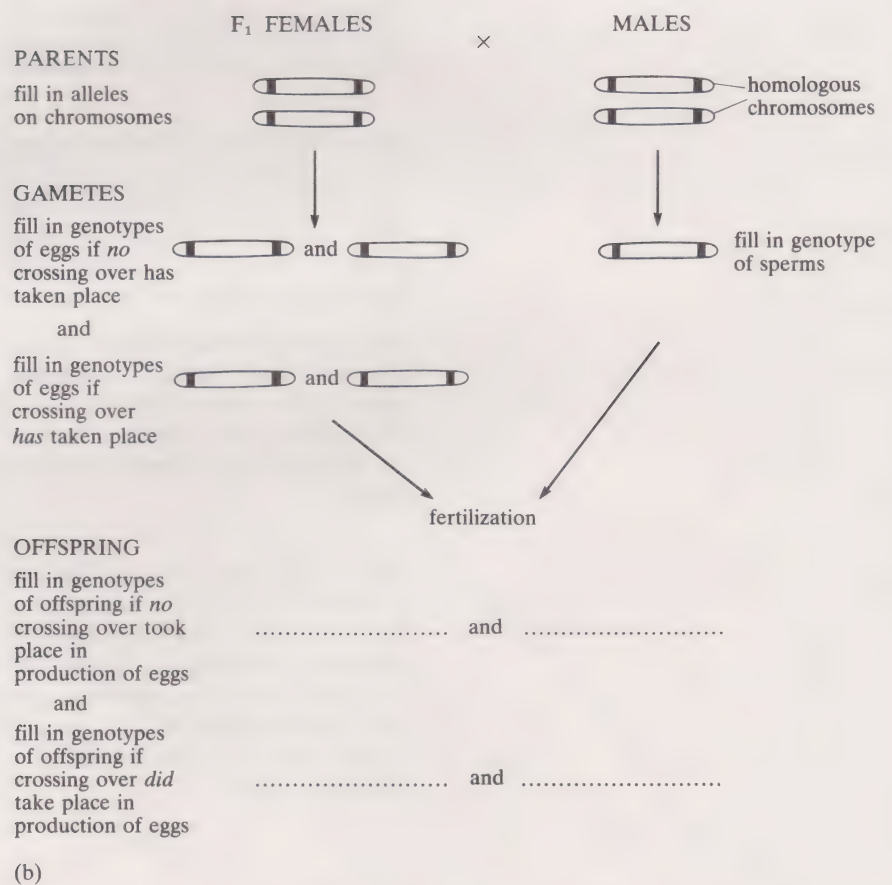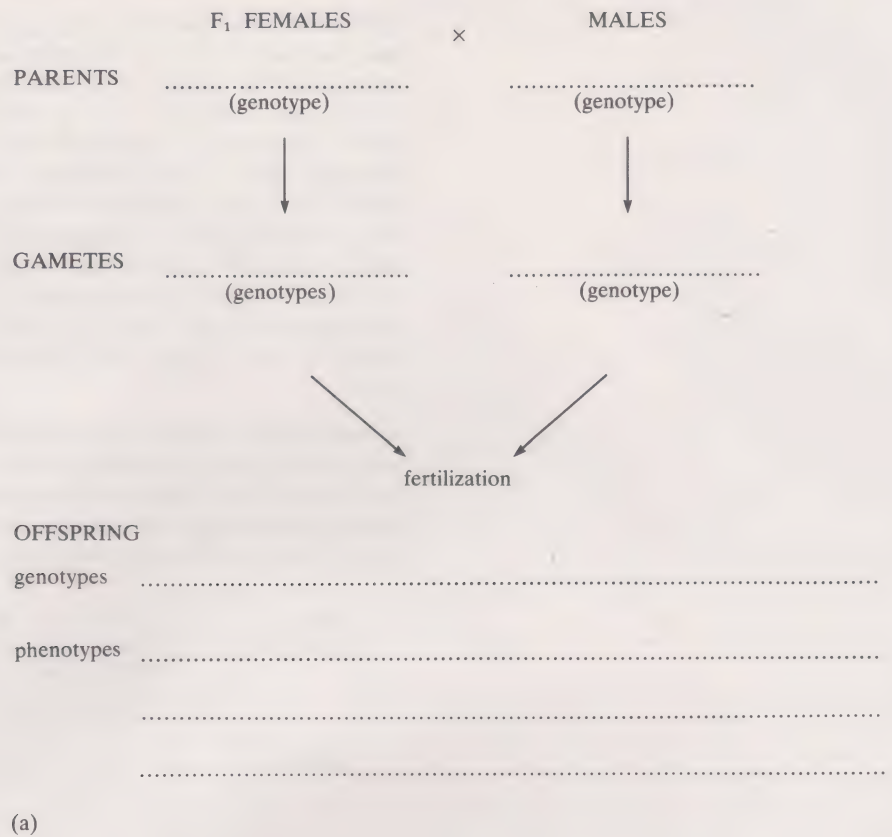
LINKAGE GROUP

F₁ FEMALES × MALES

PARENTS ....
(genotype) (genotype)

GAMETES ....
(genotypes) (genotype)

fertilization

OFFSPRING

genotypes ....

phenotypes ....

....

....

(a)

F₁ FEMALES × MALES

PARENTS
fill in alleles
on chromosomes
homologous
chromosomes

GAMETES
fill in genotypes
of eggs if *no*
crossing over has
taken place
and fill in genotype
of sperms

and

fill in genotypes
of eggs if
crossing over
has taken place
and

fertilization

OFFSPRING

fill in genotypes
of offspring if *no*
crossing over took
place in
production of eggs
.... and ....

and

fill in genotypes
of offspring if
crossing over *did*
take place in
production of eggs
.... and ....

(b)

FIGURE 38   The inheritance of wing length and eye colour in *Drosophila* under two different assumptions: (a) that the genes for wing length and eye colour are on different chromosomes; (b) that the genes for wing length and eye colour are on the same chromosome.

□ Once you have completed these Figures, work out how many flies of each phenotype you would expect to find in a group of 200 offspring flies resulting from the crosses described under the two different assumptions.

■ Figure 38a introduces nothing new. Independent assortment of the genes for wing length and for eye colour during prophase I of meiosis would lead to the females' producing four genetically different types of eggs in roughly equal numbers. The males' sperms would all be genetically identical, and hence four different genotypes would be present in the offspring generation in roughly equal numbers. Thus, among 200 such flies, one would expect approximately 50 of each of the phenotypes shown at the bottom of Figure 38a.

The outcome is very different if the genes for wing length and eye colour are on the same chromosome, for they can no longer assort independently during meiosis. This is the situation in Figure 38b. In the majority of cases, the two alleles that are on the same chromosome at the beginning of meiosis will stay together on that chromosome throughout meiosis. If, as in our example, the *W* and *e* alleles are carried on one chromosome and *w* and *E* on its homologous partner, then most of the gametes will contain one chromosome carrying either *W* and *e* or *w* and *E*. Only when crossing over has occurred during meiosis will the eggs not have one of these two genotypes. Crossing over, however, will exchange the alleles, so that a few eggs carry either *W* and *E* or *w* and *e*. Just how many such eggs there are will depend upon how often crossing over occurs; the more common is crossing over, the higher the proportion of eggs with one of these two genotypes. Without knowing how often crossing over occurs it is not possible to predict the frequency of the four genotypes in the offspring generation, but among 200 such offspring one would expect to find large and equal numbers of normal-winged, cinnabar-eyed flies and vestigial-winged, normal eyed flies. One would also expect to find small and equal numbers of vestigial-winged, cinnabar-eyed flies and normal-winged, normal-eyed flies.

Table 7 (below) shows the actual numbers of the four different phenotypes obtained in a breeding experiment.

The numbers agree closely with what is expected if the genes for wing length and eye colour are carried on the same chromosome, and are very different from the results expected if the genes were, instead, carried on different chromosomes. We therefore conclude that the genes for wing length and eye colour are carried on the same chromosome.

We can use this example to make another generalization about inheritance. We can envisage the genes of each chromosome as being linked together in a cluster, or a **linkage group** as it is also called, with one group per chromosome. Genes from one linkage group always assort independently of genes from another linkage group during meiosis because the different chromosomes on which they are carried align themselves independently at metaphase I. Genes *within* a linkage group assort together, as a unit, except when they are separated by crossing over. Note that crossing over, even if it

TABLE 7   The numbers of the four different phenotypes and genotypes in the offspring of *W w E e* female flies and *w w e e* male flies

Phenotype	Genotype	Number
normal wings, cinnabar eyes	*W w e e*	84
vestigial wings, normal eyes	*w w E e*	93
vestigial wings, cinnabar eyes	*w w e e*	12
normal wings, normal eyes	*W w E e*	11

RECOMBINATION

MITOSIS

is very infrequent, increases the genetic variability of the gametes. If there were no crossing over in the fruit-fly example, F_1 females would produce only two types of eggs, $W e$ and $w E$, and hence only two types of offspring, $W w e e$ and $w w E e$, when crossed with homozygous recessive males. With crossing over, however, F_1 females produce eggs of four different genotypes, and hence four different genotypes of offspring. The shuffling of alleles that results either from crossing over or from independent assortment is technically called **recombination**. Recombination during meiosis results in an increased genetic variability of the offspring. You know from Unit 19 that any genotypic variability that leads to phenotypic variability can have very important consequences for evolution. What these consequences are with respect to recombination is discussed in the next Unit.

SUMMARY OF SECTION 5

1   In this Section, the model of the patterns of inheritance developed in Section 4 is applied to situations involving the inheritance of more than one pair of contrasting characters.

2   Two different cases have to be considered separately: (i) where the genes for the different pairs of characters are situated on *different* pairs of homologous chromosomes; (ii) where they are situated on the *same* pair.

3   In the former case, the model accounts well for the results of breeding experiments provided it is assumed that the non-homologous chromosomes assort independently of one another during meiosis.

4   Assuming independent assortment, the model predicts that if two pure-breeding strains differing in two characters for which they are pure-breeding are crossed, then all of the F_1 generation will be phenotypically identical, manifesting the two dominant characters.

5   When individuals from the F_1 generation are crossed, the model predicts that there will be four phenotypes among the offspring:

(i) individuals with both characters dominant;

(ii) individuals with the first character dominant and the second recessive;

(iii) individuals with the first character recessive and the second dominant;

(iv) individuals with both characters recessive.

The model also predicts that these four phenotypes will be in the ratio 9 : 3 : 3 : 1. However, the random nature of independent assortment, and of the processes that determine which gametes achieve fertilization, result in actual ratios of phenotypes in breeding experiment departing somewhat from this ratio.

7   When the genes for the different pairs of contrasting characters are situated on the same pair of homologous chromosomes they do not assort independently during meiosis. They belong to the same linkage group. Genes belonging to the same linkage group remain together during meiosis, except when separated by crossing over. Crossing over thus leads to a recombination of the genes.

8   Recombination of genes by independent assortment or crossing over increases the potential for genetic variability in gamete formation. This has important evolutionary consequences.

SAQ 10   In a breeding experiment pure-breeding plants that have yellow, smooth stems are crossed with pure-breeding plants that have green, rough stems. All of the F_1 plants have yellow, smooth stems. These are then crossed with each other to produce an F_2 generation. If the genes for stem colour and texture are linked and if no crossing over occurs during meiosis, what will be the genotypes and phenotypes of the F_2 generation, and in what ratios will they occur?

Use appropriate symbols for alleles for stem colour and stem texture. (Remember that capital letters are used for dominant alleles and small letters for recessive alleles.)

SAQ 11   If the genes making up the genotype of an organism were shown to be organized into five linkage groups, how many chromosomes would you expect to find: (i) in each gamete formed by the organism; (ii) in each fertilized egg of this organism?

SAQ 12   A plant that is homozygous for the four dominant alleles A, B, Q and R is crossed with a plant that is homozygous for the four recessive alleles a, b, q and r. Each F_1 plant resulting from this cross therefore has the genotype $AaBbQqRr$. Alleles A and B are linked, as are alleles a and b. Q and R, are linked, as are q and r. A and B are not linked to Q and R, nor are a and b linked to q and r.

The genotypes of some of the gametes an F_1 plant could produce are:

(i)	$A\ B\ Q\ R$	(v)	$a\ b\ q\ r$
(ii)	$A\ b\ q\ r$	(vi)	$a\ B\ Q\ R$
(iii)	$A\ B\ q\ R$	(vii)	$a\ B\ Q\ r$
(iv)	$a\ b\ Q\ R$	(viii)	$A\ B\ q\ r$

Compare these genotypes with the genotypes of the gametes produced by the homozygous parents, and identify the F_1 gamete genotypes whose production has:
(a) not involved recombination;
(b) involved recombination by independent assortment but not by crossing over;
(c) involved recombination by crossing over.

SAQ 13   A heterozygous plant with the genotype $PpTt$ produces four kinds of gametes, PT, pt, Pt and pT in the ratio $1:1:1:1$. Another heterozygous plant of the same species has the genotype $PpQq$. This plant produces four kinds of gametes PQ, pq, Pq and pQ in the ratio $8:8:1:1$. How can these differences between the two ratios be explained?

6 MITOSIS AND GENES

This Section looks at the events that take place within a cell nucleus when cells divide during normal body growth. The Section also discusses more fully what exactly a gene is.

6.1 THE MITOTIC CYCLE

There are a few loose ends to tie up in this Unit. The first is to do with the kind of cell division that takes place when the mature organism develops from the diploid zygote that is formed when the egg is fertilized.

Remember from Section 3.2 that the nuclei of the non-reproductive cells of the body all have the same diploid complement of chromosomes. Hence, as cells divide, and a mature organism develops from the zygote, each division produces new cells that are genetically identical to their predecessors.

☐ Why could such cell division *not* proceed by meiosis?

■ Because meiosis results in the production of cells that are *not* identical to their predecessors: (i) they are haploid instead of diploid; (ii) the independent assortment of non-homologous chromosomes and crossing over produces cells that are genetically different from their predecessors.

Cell division during the growth (and indeed the repair) of an organism takes place by means of a process different from meiosis. This process is called **mitosis.** Most of you will study it further at Summer School, but we explain its main features here, once again by means of an AV sequence.

This AV sequence, 'Mitosis and meiosis compared', is on Tape 3 (Side 2, Band 4). As you listen to the tape you will need to refer to Figure 39 and Frames 11 to 16. The AV sequence should take about 20 minutes to work through.

---

11 **Mitosis : prophase**   **Meiosis : prophase I**

chromatid

centromere

nuclear envelope

---
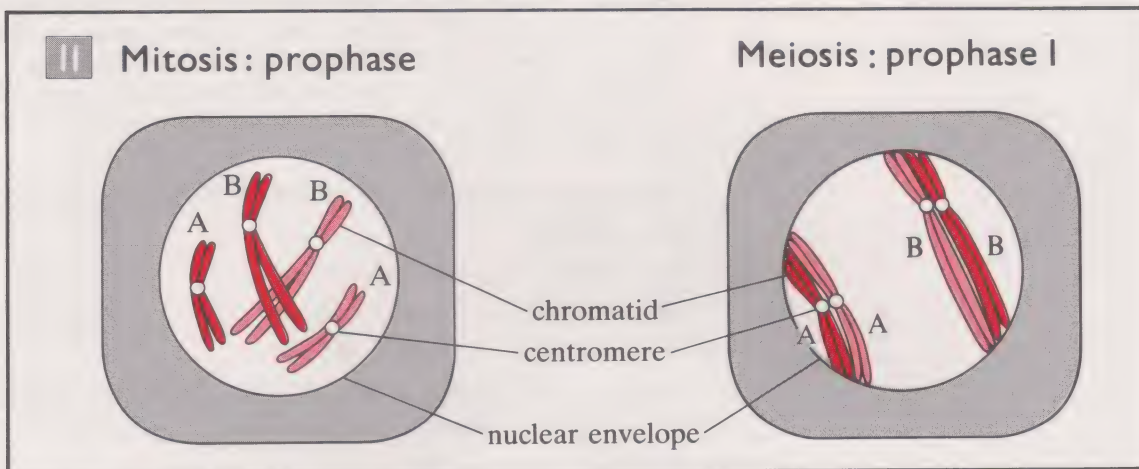
12 Differences between prophase of mitosis and prophase I of meiosis

....

....

....

....

---

13 **Mitosis : metaphase**   **Meiosis : metaphase I**
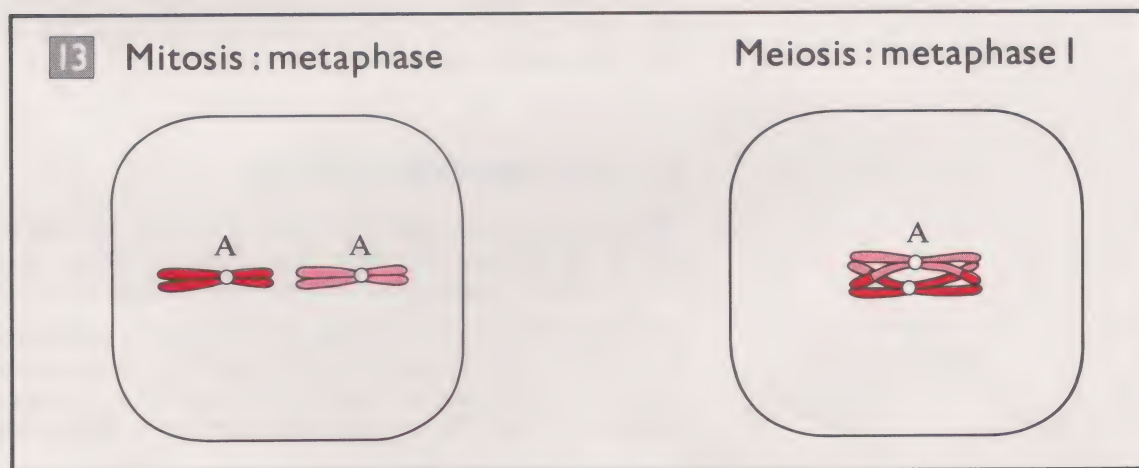
14 Differences between metaphase of mitosis and metaphase I of meiosis

....

....

....

....

15 Mitosis : anaphase   Meiosis : anaphase I
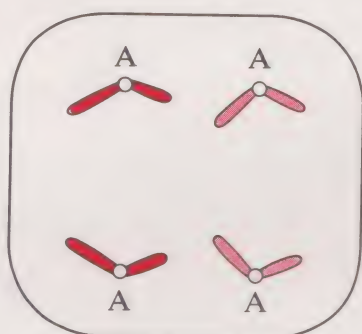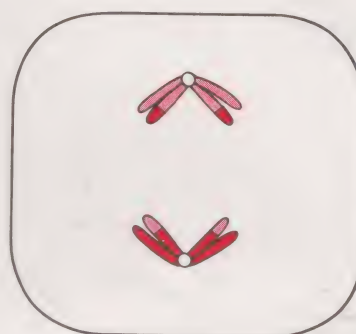
16 Mitosis : telophase   Meiosis : telophase I
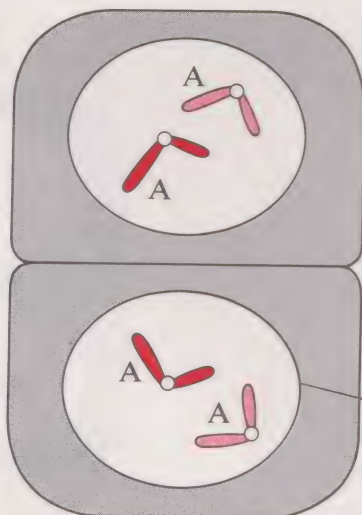
nuclear envelope

MITOSIS

DIVISION I OF MEIOSIS

Chromosomes appear divided into chromatids. Red chromatids have been inherited from the mother, pink from the father.

chromatid

centromere

nuclear envelope

prophase

chromatid

centromere

nuclear envelope

Nuclear envelope disappears and chromosomes line up around 'equator'. (Only chromosome pair A are shown.)

metaphase

The spindle draws the chromosomes to opposite ends of the cell.

anaphase

Nuclear envelopes develop and cell divides into two.

nuclear envelope

telophase

nuclear envelope

FIGURE 39 A comparison of mitosis and division I of meiosis.

You should now study Figure 39 again to help you grasp the main features of mitosis, and the main ways in which it differs from meiosis. These are:

1 Mitosis involves only one cell division. Meiosis involves two.

2 Mitosis does *not* involve pairing of homologous chromosomes. Meiosis does.

3 Mitosis produces cells whose chromosomes are exact copies of those of the parent cells. Meiosis does not.

4 Mitosis produces cells with the same number of chromosomes as the parent cells. Meiosis produces cells with half the number of chromosomes as the parent cells.

One question may have occurred to you as you read that summary. If an organism arises from a zygote by repeated mitotic division, and if mitosis produces cells with identical sets of chromosomes, why aren't the cells of the body of an animal or a plant identical to one another? In the human body for example, how can we have muscle cells, brain cells, nerve cells and skin cells that are all so different from each other when they have all arisen by mitosis from just one fertilized egg? To answer this question, you have to know more about how genes work—a topic to which we return in Unit 24.

The cell is a hive of activity during mitosis, and the machinery that underlies this activity has been and is being intensively studied. Particularly interesting is the mechanism responsible for the movement at anaphase of the chromatids towards the poles. In comparison with the many hours that the whole process of mitosis can occupy, anaphase is over in a few minutes, all of the chromatids moving at the same speed of about $1\,\mu m$ per minute. As in meiosis, the chromatids bear a number of fibres, the kinetochore fibres, sticking out from the region of the centromere. Before metaphase, the chromosomes are wafted around the cell until their kinetochore fibres become entangled in the spindle fibres. Once caught, they become lined up so that one set of kinetochore fibres are attached to the spindle fibres running from one pole, and the other set to the spindle fibres running from the other (Figure 40). The movement of the chromatids at anaphase results from three processes. First the centromere splits, with the result that the two chromatids are no longer held together. Second the kinetochore fibres start to pull their chromatids towards their respective poles. Third, the spindle fibres grow longer, pushing the two poles of the spindle farther apart, and with them the attached chromatids.
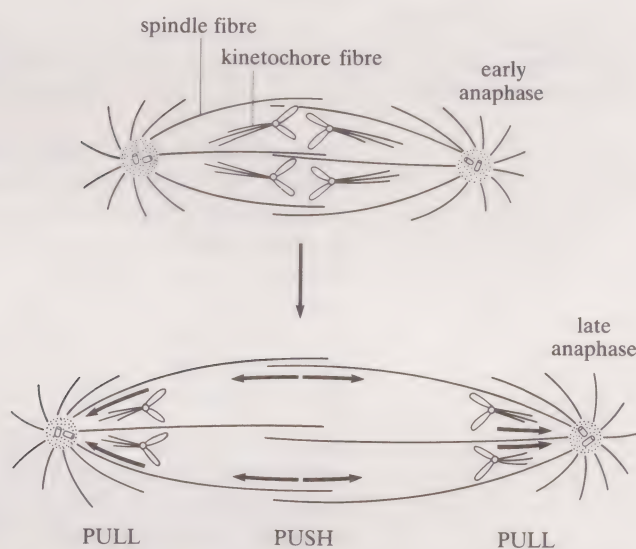
FIGURE 40 Chromatids move apart during anaphase of mitosis because of the pushing of the spindle fibres and the pulling of the kinetochore fibres.

Compared with many things that happen within the cell, mitosis in general, and anaphase in particular, is a fast-moving and energy-consuming process. Mitosis has important genetic consequences, in that it produces genetically identical 'daughter' cells. It also has important consequences in terms of the organism's energy needs, as you will discover in Unit 22.

6.2 WHAT IS A GENE?

This Unit has jumped backwards and forwards between the classical breeding experiments and modern investigations of chromosome structure and function. The model developed in Section 4 to explain the results of the breeding experiments introduced the key concept of the gene. The gene, in a breeding experiment, is the unit of material on the chromosome that is responsible for a particular phenotypic character. The gene for a character such as eye colour comes in alternative forms called alleles: there is an allele for cinnabar eyes as opposed to red ones, or for a purple maize grain as opposed to a white one. How does this definition of a gene fit in with modern knowledge of chromosome structure? Can one identify a chunk of chromosome and say: 'There is the gene for cinnabar eyes'? The answer is 'yes and no'.

It is certainly possible to produce maps showing where genes lie on chromosomes. Figure 41 shows a map of one of the chromosomes of the fruit-fly *Drosophila melanogaster*; you can see the genes for various characters strung out unevenly along its length. Furthermore, it is possible in

FIGURE 41 Genetic map of chromosome 2 of *Drosophila melanogaster*. The numbers denote the relative distance along the chromosome, as determined by measuring the frequency with which the genes are separated by crossing over, in breeding experiments.
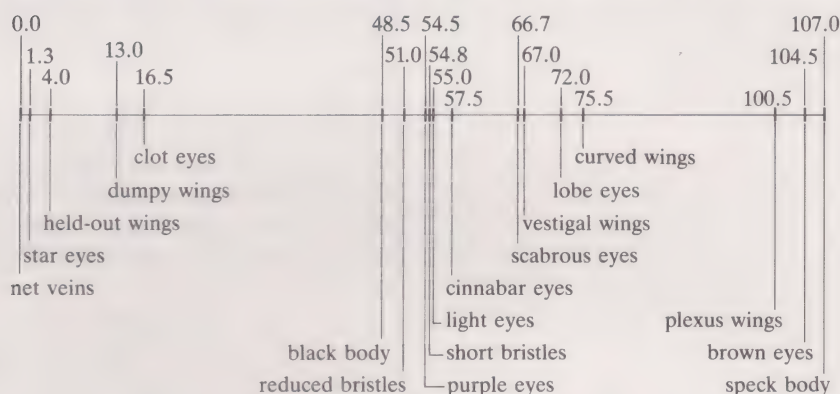
some special cases to relate some genes to quite specific parts of chromosomes. For example, the chromosomes in some of *Drosophila*'s cells are exceptionally large and have bands running across them (Figure 42). There is good evidence that each of these bands corresponds to a gene. For example, in one region on one of the chromosomes where there are 50 bands it has also been found that there are 50 genes. As mentioned in Section 3.3, each of the bands probably corresponds to a region where the chromatin loops are packed in a particular formation. If this is so, then a gene corresponds to a particular region of this sort.

However, this picture is simplistic and incomplete. It is now known that a gene on a chromosome that is responsible for a particular phenotypic character can be interrupted by lengths that do *not* code for that character: these are called 'split genes'. It is also known that certain genes can physically change their place on chromosomes, moving from one locus to another. These are known as 'transposable elements' or 'jumping genes'.

Even when it is possible to identify a chunk of chromosome as being the site of a given gene, we have still not explained how this gene functions. Why does a maize plant with purple-grain alleles develop purple grains? What is it about such an allele that ensures that the grain is indeed purple rather than white? How does the gene deliver its instructions? These are all questions for later biology Units, and particularly for Unit 24. More immediately, however, the next Unit will consider how the nature and behaviour of genes provides a source of variation on which natural selection acts.

SUMMARY OF SECTION 6

1 In mitosis, the diploid nucleus divides to produce two new nuclei that are genetically identical to itself and to each other. Unlike meiosis, mitosis consists of a single cycle of division in which single chromosomes, rather than homologous pairs, align themselves along the equator of the spindle.

right arm of
chromosome 3

X chromosome

normal mitotic
chromosomes at
the same scale

chromosome 4

region where the two
homologous chromosomes
are separated

left arm of
chromosome 3

left arm of
chromosome 2

right arm of
chromosome 2

20 μm

(a)

(b)

10 μm
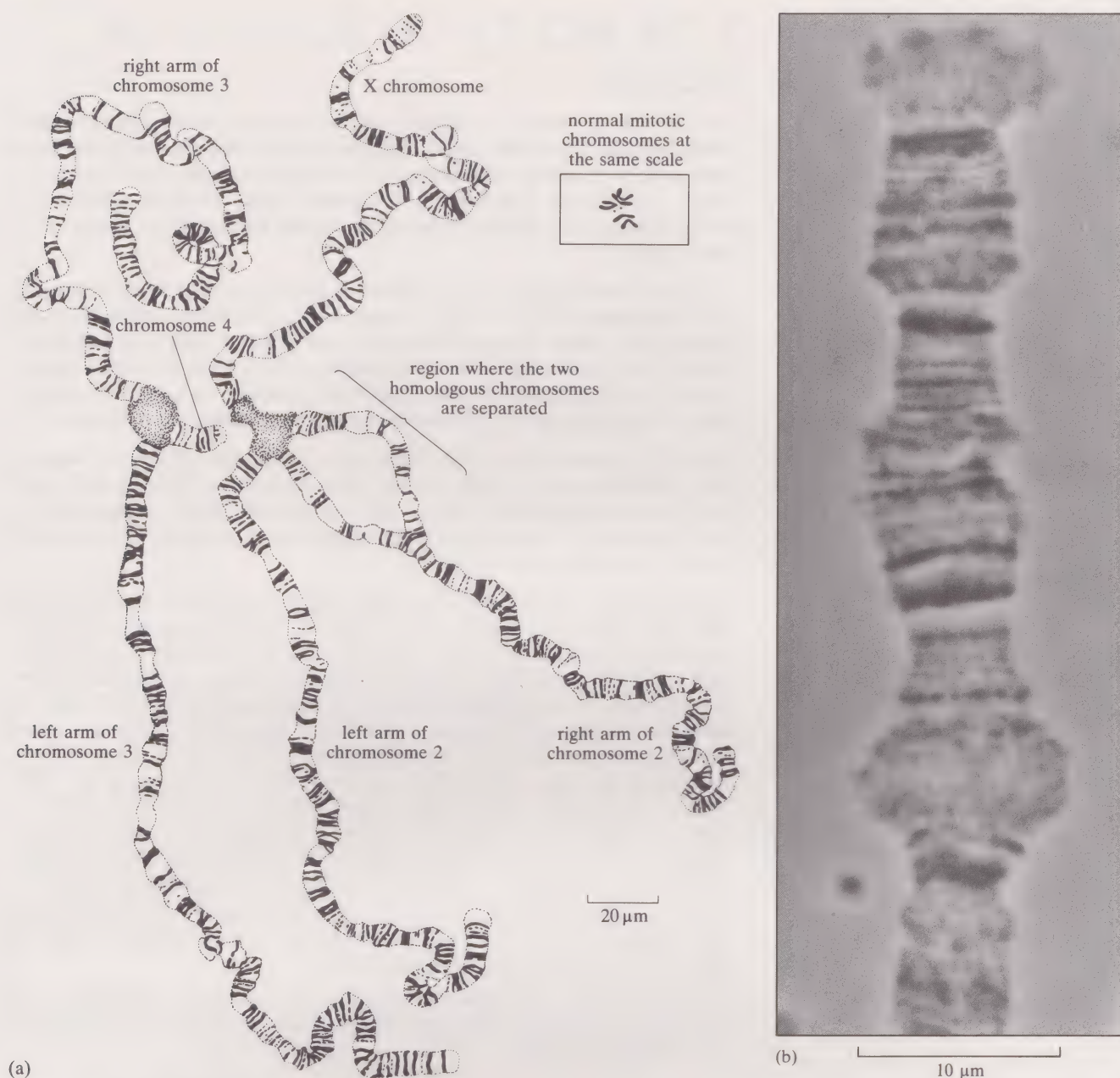
FIGURE 42 A drawing (a) and a photograph (b) of some exceptionally large chromosomes in *Drosophila melanogaster* showing the banding pattern.

Movements of the chromosomes during mitosis are achieved primarily by the kinetochore fibres, which pull the chromosomes towards the poles of the spindle, and the spindle fibres, which elongate, so forcing the chromosomes apart.

2   The gene can be defined from breeding experiments as the unit of genetic material responsible for a particular phenotypic character. Although a gene defined in this way exists as a chunk of DNA on a chromosome, the idea that 'one chunk of DNA equals one phenotypic character' is too simple.

SAQ 14   Consider an F_1 maize plant that is heterozygous for grain colour, genotype $P\,p$. Is it possible, during mitosis, to produce some daughter cells with genotype $P\,P$ and some with genotype $p\,p$? Explain your answer.

SAQ 15   (a) What are the main differences between mitosis and division I of meiosis?

(b) Compare and contrast the behaviour of homologous chromosomes at metaphase I of meiosis and at metaphase of mitosis.

7 TV NOTES: A SCHOOL OF GENES

As you read in Section 2, Mendel carried out his famous genetic experiments on peas. Since then, many different sorts of animals and plants have been used in breeding experiments in order to understand more about the basis of inheritance. The fruit-fly, *Drosophila*, which you have already met in this Unit, is one particular favourite among geneticists, as is the laboratory mouse.

You may have begun to wonder whether geneticists have studied humans, and if so, what they have found. People come in different shapes, sizes and colours; they speak different languages, they differ in their susceptibility to disease, their physiology, their biochemistry, they are either male or female, and so on. How much of this phenotypic variation is genetic in origin? What do we know about the inheritance of human phenotypic characters?

In the TV programme for this Unit, you will find the answers to some of these questions—or at least partial answers, because there is still much about human genetics that is not fully understood. In the programme, we start by visiting a school and investigating the phenotypic variation that occurs among the children there.

After you have watched the TV programme you will realize that there is at least one respect in which we have simplified the situation with respect to alleles at a given locus. So far in this text, we have considered cases where there are only two alleles of a gene available to occupy a particular locus. For example, we considered the allele for purple-grained maize, P, and the allele for white-grained maize, p. There are many cases, however, where there are not just two but several different alleles—all of them different versions of the same gene—that are potentially able to occupy the same locus on a particular chromosome. There are only two places available for them, of course, one on each chromosome of a homologous pair. To give an example from the TV programme, the ABO human blood group system is controlled by the alleles A, B and O. Individuals with genotypes AO or AA belong to blood group A; individuals with genotypes BO or BB belong to blood group B; individuals with genotype AB belong to blood group AB; and with genotype OO to blood group O.

Sometimes, as you will discover in the TV programme, the situation can be even more complicated than this, where there are dozens of different versions of the same gene.

OBJECTIVES FOR UNIT 20

After you have worked through this Unit you should be able to:

1   Explain the meaning of, and use correctly, all the terms flagged in the text.

2   Interpret simple breeding experiments in terms of Mendelian genetics. (*SAQs 1, 2, 6 and 7*)

3   Explain the implications of the various stages of meiosis for the distribution of chromosomal material and genes among cells and within nuclei. (*SAQs 3, 4, 5, 8, 9, 10, 11, 12 and 13*)

4   Explain the implications of the various stages of mitosis for the distribution of chromosomal material and genes among cells and within nuclei. (*SAQ 14*)

5   Summarize the main differences between meiosis and mitosis. (*SAQ 15*)

ITQ ANSWERS AND COMMENTS

ITQ 1 Cob A contains both purple and white grains. White grains had vanished in the F_1 generation, but have now reappeared in the F_2. Thus, even though there were no white grains in the first generation of offspring, the factors responsible for the inheritance of white grain colour must still have been present and handed on to the second generation of offspring.

ITQ 2 Your results should have given you a ratio of purple grains to white grains of around 3 : 1.

Exactly how many purple and white grains a cob contains varies from one cob to another. Values we have found in cobs that we have looked at are shown in Table 8. See how your own values compare with them. For each cob in Table 8 the ratio of purple to white grains, to the nearest whole numbers, is 3 : 1.

TABLE 8 Numbers of purple and white grains in maize cobs similar to cob A in the Kit

Total no. of grains	No. of purple grains	No. of white grains	Ratio of purple to white grains
348	249	99	2.52:1
702	541	161	3.36:1
566	437	129	3.39:1
424	310	114	2.72:1
679	499	180	2.77:1
534	388	146	2.66:1
391	284	107	2.65:1
495	378	117	3.23:1

ITQ 3 Table 9 is the completed Table 3.

TABLE 9 For ITQ 3

The fertilized egg in box 1 is homozygous for the dominant character, so will have purple grains. The fertilized eggs in boxes 2 and 3 are heterozygous, and will therefore have purple grains. (Remember that it does not matter which parent contributes P and which contributes p, hence the genotype Pp produces the same phenotype as does genotype pP.) The fertilized egg in box 4 is homozygous for the recessive character. It will have white grains. Thus three of the boxes give purple grains; one of them gives white.

ITQ 4 Figure 43 (overleaf) is the completed Figure 32. All the gametes produced by the plants with purple, smooth grains (a) will have the P and S alleles, while all the gametes produced by the parents with white, wrinkled grains (b) will have the p and s alleles.

ITQ 5 Figure 44 (on p. 55) is the completed Figure 35.

ITQ 6 Table 10 (on p. 55) is the completed Table 4. It shows the genotypes and phenotypes of the offspring of parents heterozygous for the same pair of contrasting characters: grain colour and coat texture.

ITQ 7 You should have found that the ratio of the four phenotypes in your maize cob B was roughly 9 : 3 : 3 : 1. Table 11 shows the results for similar cobs. Note the considerable variation both in the total number of grains present in each cob and in the actual ratio of the four phenotypes.

TABLE 11 Numbers of grains of four different phenotypes in maize cobs similar to cob B in the Kit (for ITQ 7)

Cob no.	Purple, smooth	Purple, wrinkled	White, smooth	White, wrinkled	Total
1	190	51	78	29	348
2	407	139	118	38	702
3	386	117	124	40	667
4	204	53	78	17	352
5	307	101	92	33	533

ITQ 8 Figures 45a and b (on p. 56) are the completed Figures 38a and b, respectively.
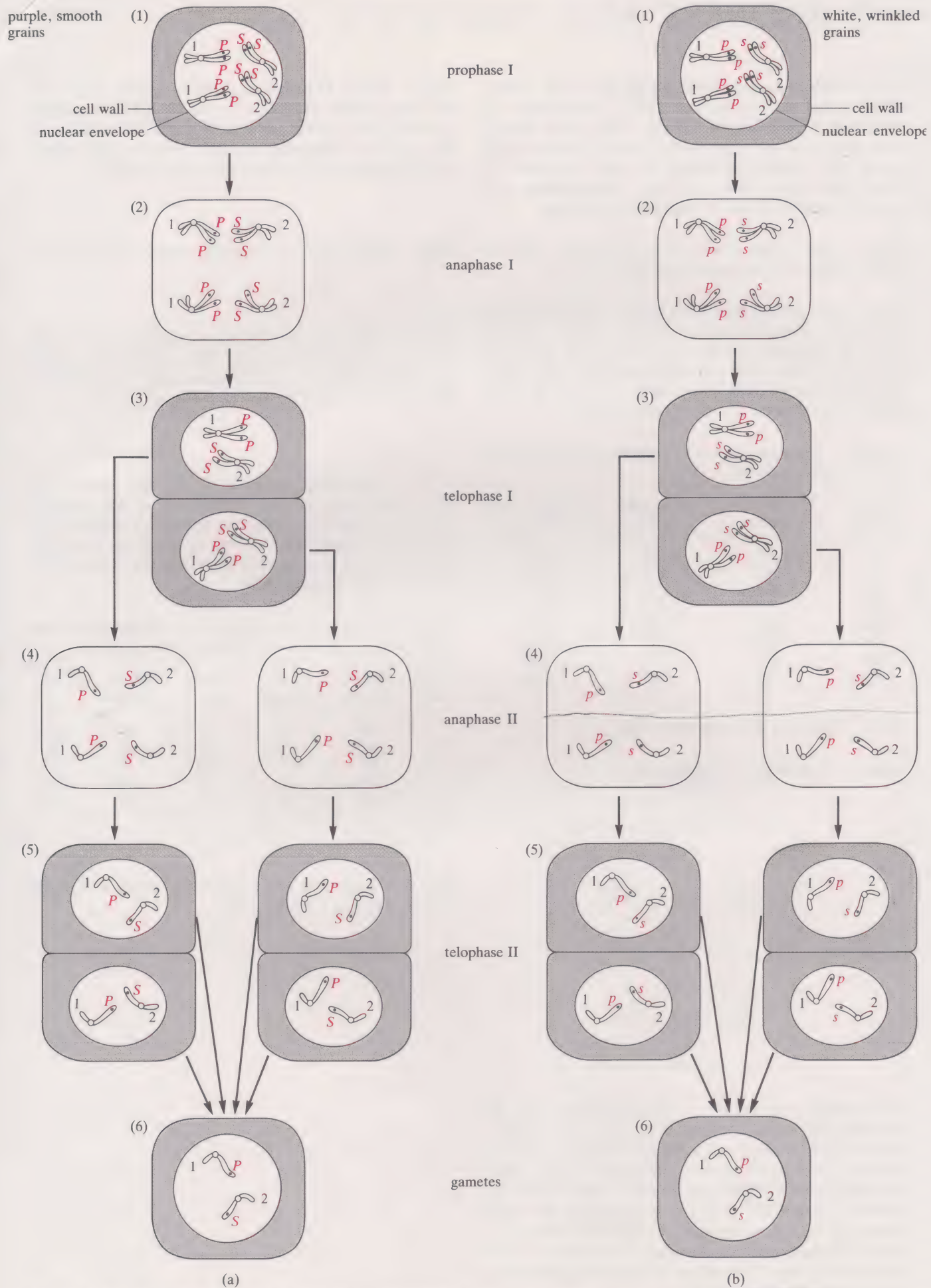
FIGURE 43 The fate of two pairs of genes on different pairs of homologous chromosomes during meiosis (for ITQ 4).
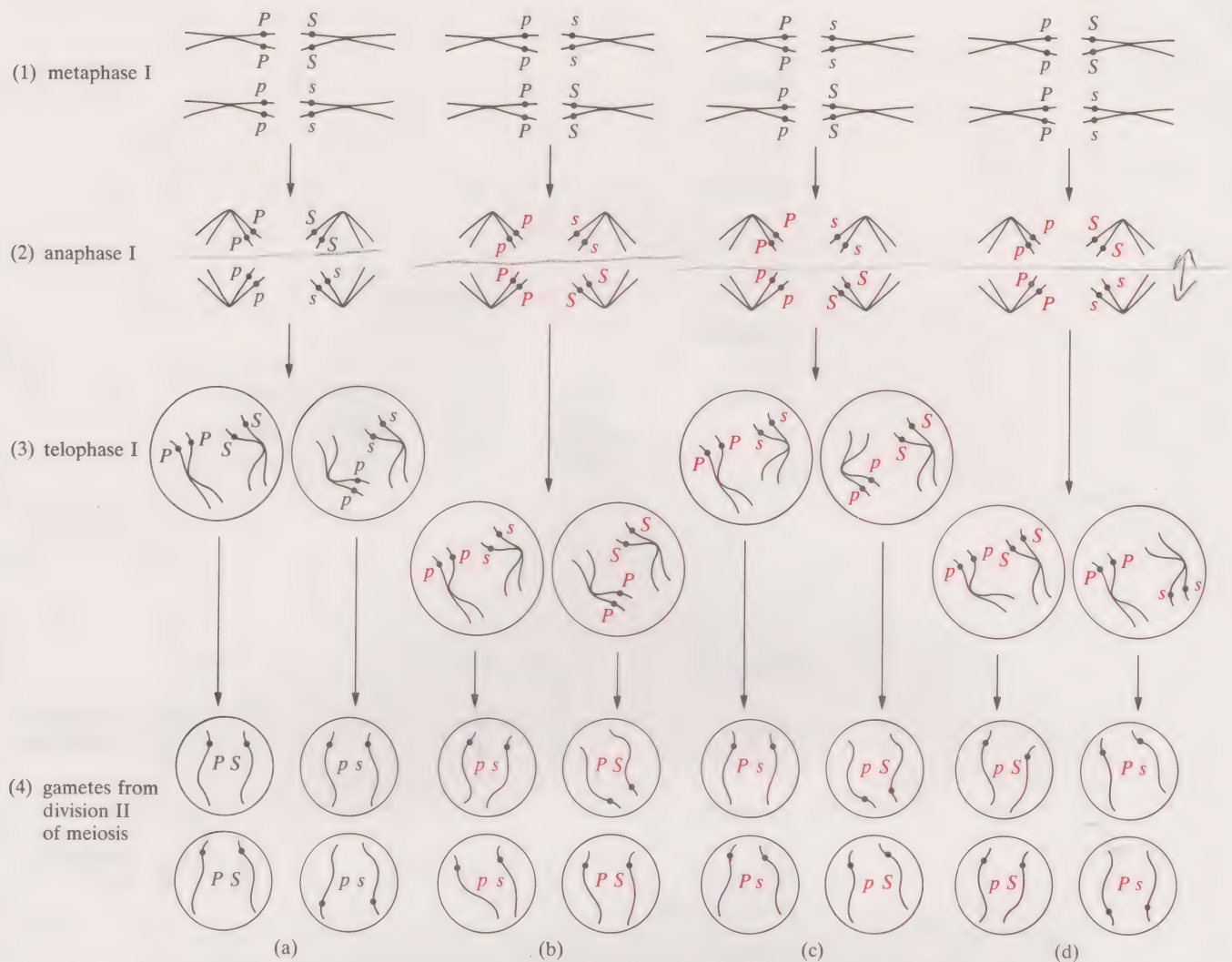
FIGURE 44 The genotypes of gametes resulting from each of the four different arrangements of chromosomes at metaphase I of meiosis (for ITQ 5).

	genotypes of female gametes			
	$P\,S$	$P\,s$	$p\,S$	$p\,s$
$P\,S$	$P\,P\,S\,S$ purple, smooth 1	$P\,P\,S\,s$ purple, smooth 2	$P\,p\,S\,S$ purple, smooth 3	$P\,p\,S\,s$ purple, smooth 4
$P\,s$	$P\,P\,s\,S$ purple, smooth 5	$P\,P\,s\,s$ purple, wrinkled 6	$P\,p\,s\,S$ purple, smooth 7	$P\,p\,s\,s$ purple, wrinkled 8
$p\,S$	$p\,P\,S\,S$ purple, smooth 9	$p\,P\,S\,s$ purple, smooth 10	$p\,p\,S\,S$ white, smooth 11	$p\,p\,S\,s$ white, smooth 12
$p\,s$	$p\,P\,s\,S$ purple, smooth 13	$p\,P\,s\,s$ purple, wrinkled 14	$p\,p\,s\,S$ white, smooth 15	$p\,p\,s\,s$ white, wrinkled 16

genotypes of male gametes

TABLE 10
F_2 genotypes and phenotypes (for ITQ 6)

55

F₁ FEMALES   MALES

×

PARENTS   *W w e E*   *w w e e*

GAMETES   *We   WE   we   wE*   *we*

fertilization

OFFSPRING

genotypes   *W w e e*   *W w E e*   *w w e e*   *w w E e*
phenotypes   normal   normal   vestigial   vestigial
                   wings,   wings,   wings,   wings,
                   cinnabar   normal   cinnabar   normal
                   eyes   eyes   eyes   eyes

(a)

PARENTS   F₁ FEMALES   ×   MALES

alleles
on chromosomes

GAMETES

genotypes of
eggs if *no*   *W*   *e*   and   *w*   *E*   *w*   *e*   genotype of
crossing over   sperms
has taken place

genotypes of
eggs if   *w*   *e*   and   *W*   *E*
crossing over
has taken place

fertilization

OFFSPRING

genotypes of
offspring if *no* crossing   *W w e e*   and   *w w E e*
over took place in
production of eggs

genotypes of
offspring if crossing   *w w e e*   and   *W w E e*
over *did* take place in
production of eggs

(b)

FIGURE 45   The inheritance of wing length and eye colour under two different assumptions (for ITQ 8).

SAQ ANSWERS AND COMMENTS

SAQ 1 Options 1 and 4 could have involved pure-breeding flies.

In option 1, the 300 white-eyed parents could have been pure-breeding, as could the 300 red-eyed parents. You are not told what offspring result when the white-eyed parental strain are bred with each other, nor what offspring result when the red-eyed parental strain are, so you have no direct evidence that either is pure-breeding. However, they both certainly could be pure-breeding and, indeed, the situation is directly analogous to the maize example in Section 2.1.

Option 2 cannot have involved pure-breeding parents since they have produced two kinds of offspring.

Option 3 cannot have involved pure-breeding flies since half of the offspring are unlike the parents, all of which were white-eyed.

In option 4, all of the large number of crosses between the white-eyed flies produce white-eyed offspring, and this is the result you would expect to find if the parental flies are pure-breeding.

SAQ 2 The results of Experiment 1 differ from those obtained by Mendel because in this experiment the character that vanishes in the first offspring generation then reappears in about three-quarters of the individuals in the second generation. This is the other way round from Mendel's results where it was the character that persisted in the first offspring generation that reappeared in about three-quarters of the second generation.

The results of Experiment 2 differ from those obtained by Mendel because the colour of the first generation of offspring is intermediate between those of the parents. Mendel found instead that one parental character persisted and the other vanished.

The results of Experiment 3 differ from those obtained by Mendel in that the outcome was different depending upon which plant provided the eggs and which the pollen.

SAQ 3 At the beginning of meiosis, each of the chromosomes has already divided into two chromatids, so there is double the amount of chromosomal material present in the cell. Two successive divisions, each halving the amount of chromosomal material, are therefore needed to produce the haploid condition.

SAQ 4 (i) Crossing over occurs between chromosomes that are homologous with one another; it does not occur between non-homologous chromosomes. (ii) The two chromatids involved in a crossing over come from different chromosomes, not from the same chromosome. (iii) Crossing over occurs during prophase I and not prophase II (see Figure 10).

SAQ 5 In division II of meiosis homologous chromosomes are not present in the same cell; they have become separated during division I (see Figure 10).

SAQ 6 The genotypes of the offspring will be PP and Pp. All will have purple grains. The PP and Pp geno-

types will be present in a ratio of $1:1$. The explanation for this is as follows.

The pure-breeding purple-grained plants will produce gametes that all carry the P allele. The heterozygous Pp plants will produce gametes, half of which carry the P allele and half the p. Using symbols, the cross is as follows.

parents	PP	$\times$	Pp
gametes	P	P and p
F_1		PP and Pp

Half of the offspring will have the genotype PP and half Pp. All the offspring will therefore have purple grains, since they all carry at least one dominant allele (P).

SAQ 7 The genotypes of the offspring will be pp (which will have white grains) and pP (which will have purple grains). The two genotypes (and phenotypes) will be present in the ratio of $1:1$. The explanation for this is given below.

The pure-breeding white-grained plants will produce gametes that all carry the p allele. The heterozygous Pp plants will produce gametes, half of which carry the P allele and half the p. The cross can therefore be represented as follows.

parents	pp	$\times$	Pp
gametes	p	P and p
F_1		pP and pp

Half of the offspring will have the genotype pP and half pp. Half will therefore have purple grains and half will have white. (*Note*: When writing down a heterozygous genotype, it is usual to write the capital letter first and the lower case letter second. So here the conventional way of writing the heterozygous genotype is Pp.)

SAQ 8 Figure 46 is the completed Figure 29. There is no other answer (assuming that a mutation has not occurred). The chromosome inherited from one parent carrying the P allele has replicated to become two identical chromatids, each carrying the same allele. Similarly, the chromosome inherited from the other parent carrying the p allele has replicated to become two chromatids, each carrying the p allele.
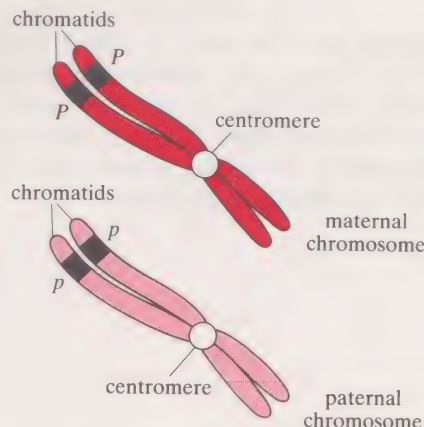
FIGURE 46 The chromatids of one pair of homologous chromosomes at the start of meiosis (for SAQ 8).

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SAQ 9 Yes. The arrangement could be different as a result of crossing over between chromatids from homologous chromosomes. As a result, one of each pair of chromatids could carry a P allele and the other member of the pair, a p allele.

SAQ 10 There will be three genotypes: $YYSS$, $YySs$ and $yyss$, in the ratio 1 : 2 : 1, respectively. The corresponding phenotypes will be yellow, smooth-stemmed and green, rough-stemmed, in a 3 : 1 ratio. The explanation for this answer is as follows.

Since all the F_1 offspring have yellow, smooth stems, we know that yellow is dominant to green and smooth is dominant to rough. So the appropriate letters for the four alleles are:

Y = yellow, y = green, S = smooth, s = rough.

Because the genes for stem colour and texture are linked and because no crossing over occurs, then they will always assort together; that is, the Y allele will always go with the S allele, and the y with the s. You would therefore expect two genes that are linked so completely that no crossing over occurs between them, to be inherited in the same way as a single gene. The breeding crosses can be depicted as follows.

parental generation	$YYSS$ (yellow, smooth)	×	$yyss$ (green, rough)
gametes	YS		ys
F_1		$YySs$	
gametes		YS and ys in equal numbers	

		female gametes	
		YS	ys
F_2 male gametes	YS	$YYSS$ yellow, smooth	$YySs$ yellow, smooth
	ys	$yYsS$ yellow, smooth	$yyss$ green, rough

The genotypic ratio in the F_2 will therefore be: $1YYSS : 2YySs : 1yyss$, and the phenotypic ratio will be three yellow, smooth-stemmed plants to one green, rough-stemmed plant. This 3 : 1 ratio is just what you would expect if the genes for stem colour and stem texture behave as though they are one.

SAQ 11 Genes that are in the same linkage group are on the same chromosome, so you would expect to find (i) five chromosomes in each gamete and (ii) ten chromosomes (i.e. five pairs) in each fertilized egg.

SAQ 12 (a) (i) and (v); (b) (iv) and (viii); (c) (ii), (iii), (vi) and (vii).

(a) Recombination means the appearance of new arrangements of genes through either independent assortment or crossing over, or both. In (i) and (v) there is no new arrangement; therefore there has been no recombination.

(b) Recombination has occurred in all but (i) and (v). Of the six remaining genotypes, in only (iv) and (viii) is there no separation of linked alleles.

(c) The genotypes (ii), (iii), (vi) and (vii) all show evidence of separation of one or more linked genes.

SAQ 13 In the first plant the genes are on different pairs of homologous chromosomes with P and p on one pair and T and t on another pair. This means that at gamete formation the genes assort independently from each other.

In the second plant the genes must be linked on the same chromosome. This means that at gamete formation P and Q and p and q will stay together more often than they separate. Only when the linkage of P and Q and p and q is broken by crossing over will the gametes Pq and pQ be produced. As this is a rare event, most of the gametes will be PQ and pq, with only a small proportion of Pq and pQ; hence the 8 : 8 : 1 : 1 ratio.

SAQ 14 No, it will not be possible. Each chromosome carrying the P allele will pass on a chromosome with a P allele to both daughter cells. Similarly, each chromosome carrying the p allele will pass on a chromosome with a p allele to both daughter cells. Hence, all daughter cells arising from mitosis will have the genotype Pp.

SAQ 15 (a) (i) Homologous chromosomes do not pair in mitosis; they do in division I of meiosis. (ii) Mitosis produces exact copies of the chromosomes in the parent cell; meiosis does not. (iii) At the end of mitosis, each nucleus contains the diploid number of undivided chromosomes; at the end of division I of meiosis, each nucleus contains the haploid number of chromosomes, and each chromosome is divided into two chromatids.

(b) At metaphase I of meiosis, pairs of homologous chromosomes will be lined up opposite one another along the equator of the spindle. By contrast, at metaphase of mitosis, the homologous chromosomes will not be opposite one another, although both chromosomes from each pair will be aligned along the equator.

ACKNOWLEDGEMENTS

INDEX FOR UNIT 20

Character	One form of character	Alternative form of character
seed shape	round	wrinkled
seed colour	yellow	green
pod shape	inflated	constricted
pod colour	green	yellow
flower colour	purple	white
flower position	along stem	at tip
stem length	long	short

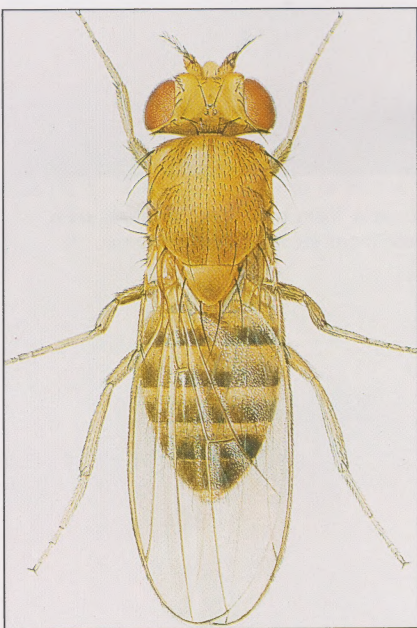PLATE 11   Pairs of contrasting characters in peas, investigated in Mendel's experiments.

PLATE 12   The fruit-fly *Drosophila melanogaster*.